



# AOGD BULLETIN

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**AOGD Theme 2018-19**  
**Empowering Providers:**  
**Enhancing Women's Health**

**Issue: Current Update**  
**Preconceptional Counseling & Prenatal Diagnosis**  
**Social Gynecology**



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# President's Message



Dear AOGD members

Greetings !

As I relinquish office after serving as President of AOGD for one year, I have a great sense of contentment that we could contribute towards the goals of AOGD mentioned below.

We started our tenure with the theme of “Empowering health care providers: Enhancing women’s health”. So at the beginning we had a motivational talk “Healing the Healers” by Dr Mohit Gupta on World Health Day dedicated to health of the doctors. A number of CMEs, Public awareness programs, and walks for creating awareness about thalassemia, environment pollution and prevention of female feticide - Beti Bachao & Beti Padhao were organised.

The 40<sup>th</sup> Annual conference of AOGD was successfully held at IHC with theme “Updating Knowledge: Enhancing Competencies.” It was inaugurated by Dr S Venkatesh- DGHS, Dr R Garg- Director LHMC and Dr Jaideep Malhotra- President FOGSI. There were six very well attended workshops organised by different institutions.

We celebrated International Women’s Day on 7<sup>th</sup> March where a motivational talk on “Nurturing the inner beauty” was delivered by Dr. Mohit Gupta followed by Yoga Session and ended with Zumba.

Continuing with the academic endeavours, Medical Education Committee of FOGSI and AOGD organized the “FOGSI FORCE Rajdhani- PG Academic programme” on 5<sup>th</sup> & 6<sup>th</sup> May 2018 and 26<sup>th</sup> & 27<sup>th</sup> March 2019 at Swarn Jayanti Auditorium, LHMC. It was aimed at teaching and updating postgraduate students to prepare them to face the examination confidently. Each Pg program was attended by around 200 students and 70 faculty. Streaming of a lecture and a case presentation was done through NMCN network to all medical colleges on 27<sup>th</sup> March.

This, last issue of Bulletin focuses on Social Obstetrics and Preconceptional Counselling & Prenatal Diagnosis which has a very important role in day to day practice and I hope you all will be benefitted after reading it. I hope, the monthly bulletin with dedicated themes were informative and you will like to preserve them for future references. These bulletin are also available on AOGD website.

The monthly clinical meeting and GBM was held at LHMC on 5<sup>th</sup> April where interesting rare cases were presented. At the end, AOGD secretariat was handed over to AIIMS. Congratulations Dr. Sunesh Kumar for taking over as President AOGD, 2019-2020. We wish him and all other office bearers the very best.

I take this opportunity to thank the Patrons, Advisors, Executive committee members and my colleagues for their unconditional support that has helped us to complete our term with a sense of satisfaction. This could not have been possible without the untiring efforts of AOGD team at Lady Hardinge Medical College. Thank you all for giving me this opportunity and wonderful experience of serving this prestigious Association of Obstetrician & Gynaecologists’ of Delhi as its President.

Long Live AOGD !  
Warm Regards,

Dr Abha Singh  
President AOGD (2018-19)

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# Secretary's Message



As we pen down the Secretary's message for the April Issue we come to the end of our tenure.

We at Lady Hardinge look back with a feel of satisfaction and pride at a very successful year of holding the AOGD Secretariat. We thank our Patrons, Advisors and the Executive Committee for their support and guidance throughout. With the active participation and enthusiasm of the subcommittee chairpersons the Association could hold number of CME'S, Public awareness programmes and workshops.

The Postgraduate teaching modules held by the infertility committee were a super success. The FOGSI Force Rajdhani for the postgraduates held twice in the last year were well appreciated and attended by the students from all over the country. Annual conference of AOGD an event eagerly awaited by almost all members was a runaway success.

AOGD was well represented at FOGSI with members earning laurels and prizes. The secretariat has been handed over to AIIMS with Dr Sunesh as President and Dr Vatsala as Secretary. We wish them and their team a very best and lots of good wishes for the year to come.

We congratulate the new subcommittee chairpersons and wish them success. A new subcommittee has been formed on Quality Assurance in Obstetrics and Gynecology.

I thank my core team from Lady Hardinge for working tirelessly the whole year and supporting in all endeavours. The editorial team had worked hard to bring out the monthly bulletins updating you with the latest evidence. The latest issue is here worth a read.

As we hand over the baton to the new team we thank all AOGD members for their unconditional support, participation and encouragement throughout the year.

Thanks to all

Dr Kiran Aggarwal  
Secretary AOGD (2018-19)

## Monthly Clinical Meeting

Monthly Clinical Meet will be held at Apollo Hospital, New Delhi on Friday, 26<sup>th</sup> April, 2019  
from 04:00pm to 05:00pm.

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## Editorial Team's Message



**Dr Ratna Biswas**  
Editor



**Dr Pikee Saxena**



**Dr Sharda Patra**  
Co-Editors



**Dr Swati Agrawal**

Hello Friends,

It is a pleasure for us to bring out the April Issue of the AOGD Bulletin which is incidentally the last bulletin under our editorial direction.

The themes of this issue are “Preconceptional Counseling & Prenatal Diagnosis” and “Social Gynecology”

The Standard of Care section in Obstetrics deals with Preconception and Prenatal Carrier Screening for Common Genetic Disorders like Thalassemia, Spinal Muscular Atrophy and Fragile X Syndrome. These are common single gene disorder which causes significant disabilities in the affected offspring, hence their early diagnosis will allow pregnancy termination. Recent advances focuses on Next Generation Sequencing in Obstetrics whereas Controversy section raises the question whether Non-Invasive Prenatal Testing (NIPT) can replace Biochemical Screening ? Case Approach on Non Immune Hydrops Fetalis (NIHF) gives guidance on how to establish the etiological diagnosis of NIHF which is a pertinent concern as it helps in risk assessment for recurrence in subsequent pregnancies.

Social Gynecology section has discussed the Management of Survivor's of Sexual Violence under the Standard of Care section. The gynecologist has the dual role of giving medical care including prophylaxis for HIV, Immunoglobulin for HbsAg and Emergency Contraception and collecting and preserving evidence for providing legal aid and justice to the victim.

Recent advances in Menstrual Hygiene has discussed the wide variety of disposable pads and re-useable cloth pads and other devices like the menstrual cups and tampons. With the burgeoning population of India the amount of menstrual waste generated is enormous and addressing the issue of appropriate waste disposal cannot be over emphasized. Indigenous clay and mud incinerators devised by women in Gujarat's village has brought forth low cost and safe menstrual waste disposal methods.

The best ways to “Approach Psychosexual Issues in Adolescents” have been addressed in controversy section. Adolescent age is the time when sexual identity is established and addressing issues on the gender identity and sexual practices in the adolescents is a very delicate situation. Sexual abuse and psychological disturbances including suicidal tendency is not uncommon and has to be dealt in a very sensitive manner. Counseling and support by doctors, social workers, family and friends is essential components of medical care.

Case approach to “Domestic Violence” is the last article which has defined what is domestic violence and how to examine and manage women with such problems.

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The motivational article on “Think before you Think” urges us to be constructive in our thoughts and channelize them to create positivity

Journal scan has brought forth recent literature on Expanded Carrier Screening and Menstrual Hygiene and Menstrual Waste Disposal. It calls for an interesting reading. The maze of knowledge-crossword and pictorial quiz is very mind tickling and must be attempted. Proceedings of clinical meeting is a must read section.

We thank our esteemed authors for their invaluable contributions. We are extremely grateful to our learned readers for their constant support and appreciation which has motivated us to work hard and bring out the best. We congratulate the AIIMS team who will take over from where we leave and will continue to bring forth these knowledge packed high quality periodicals.

Wishing all a Happy Reading!!

Editorial Team

# Preconception and Prenatal Screening for Common Genetic Disorders in India



Dr Seema Thakur

Seema Thakur<sup>1</sup>, Shubhnita Singh<sup>2</sup><sup>1</sup>Sr. Consultant, Genetic & Fetal Diagnosis, Fortis Hospital, Delhi- NCR, Apollo Hospital, Delhi & Rainbow Children Hospital, Delhi<sup>2</sup>Fellow, Genetic & Fetal Diagnosis, Fortis Hospital, Delhi

## Abstract

Prenatal carrier screening is done to identify asymptomatic individuals, so that risk of carrying an affected child with genetic disorder can be detected. The target population is couples who are pregnant or planning to become pregnant. Screening could be done for common single gene disorders or can be done for multiple disorders simultaneously by Next Generation Sequencing(NGS). India being a huge country, carrier screening is a health priority. In this article we discuss carrier testing for single gene disorders.

## Introduction

Carrier screening is for prevention of genetic disorders in fetus by detection of carrier status of prospective parents. Carriers are themselves healthy but are at risk of a child with a serious genetic disorder if their partner is also the carrier of same genetic disorder.

Each individual is a carrier of between 0-7 severe childhood recessive conditions. Because of the recessive pattern of inheritance, most carrier couples have no family history suggestive of the disorder and therefore become aware of their reproductive risk after birth of affected child. It is estimated that 5.3% of newborns will suffer from a genetic disorder, when followed up until the age of 25 years.<sup>1</sup>

## Carrier screening in India: Need of the hour

The current population of India is around 1,36 billion, based on the latest United Nations estimates. India population is equivalent to 17.74% of the total world population. India ranks number 2 in the list of countries by population. About 50% of Indian population is <25 years of age and about 51 births happen every minute.

There is a high prevalence of genetic disorders in India due to very large population and high birth rate and consanguineous marriage favoured in many communities. Also our society favours endogamy.

Table: 1 shows an estimated number of births of genetic disorders / year in India. About 5 lakhs children are born with malformations. There is a birth of about 21,000 children with Down syndrome and 9000 children with thalassemia every year<sup>2,3</sup>

Hence, prevention of genetic disorders is a major focus of preventive fetal medicine.

## Criterion for Prenatal Carrier Screening<sup>4</sup>

1. The natural history of the disorder should be well understood and should severely impair the health of an affected offspring.
2. There should be a high frequency of carriers in the screened population.
3. Technically valid screening methods should be available.
4. The genotypic and phenotypic correlations should be predictable and strong.
5. Prenatal diagnosis and intervention should be valid reproductive options.

## ACOG and ACMG Joint statement for carrier screen Guidelines for obstetric care provider-2017<sup>5</sup>

- Threshold- carrier frequency for condition-1 in 100
- Should cause Physical/Mental handicap
- No adult onset disorders should be tested

In this article we would discuss, screening for single gene disorders.

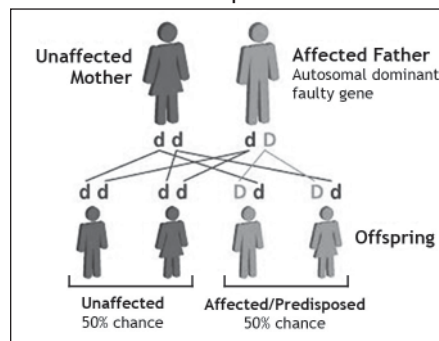
## SINGLE GENE DISORDERS

Single gene can be inherited in different patterns as mentioned below:

- Autosomal Dominant
- Autosomal Recessive
- Sex-linked/Sex-limited
- X-linked recessive/dominant

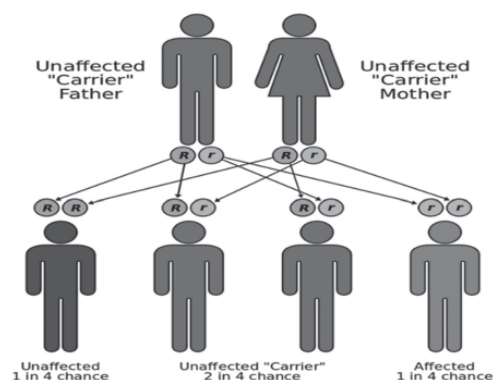
In autosomal dominant Inheritance there is-

- 50% risk of affected child with each pregnancy
- Males and females are equally affected
- Often variable expression with reduced penetrance



In autosomal recessive inheritance, both parents are carriers and

- 25% risk of affected child with each pregnancy
- Males and females are equally affected



### When should carrier screening be done?

Carrier screening is best done preconception, before IVF procedure so that there is enough time for genetic counselling.. This can also be done in early pregnancy.

### Who should get carrier screening?

Carrier screening for single gene disorders can be done universally or can be targeted in those with history of genetic disorder or at risk of genetic disorder.

- Universal/ Panethnic
- Targeted/Ethnicity based

### What are the tests for carrier screening?

Tests for Carrier screening can be broadly divided into two types:

- A. Screening for common genetic disorders
- B. Expanded carrier screening by NGS
  - A. Common genetic disorders for carrier screening in Indian population would include:
    1. Thalassemia/Haemoglobinopathy
    2. Spinal muscular atrophy
    3. Fragile X syndrome

## Hemoglobinopathies <sup>6,7</sup>

$\beta$ -thalassemia is the commonest single-gene disorder characterized by severe transfusion dependent anemia. Prevalence in India is 1.2/1,000 live births. It is estimated that 10% of the total world thalassemics are born in India every year.

In the Indian population the prevalence of  $\beta$ -thalassemia carriers is 3-4%. High-risk ethnic groups would comprise Sindhis, Kutchis, Lohanas, Punjabis, few Muslim groups as well as few tribal populations. (5-17%).

The aim of prenatal hemoglobinopathy screening is to detect and counsel asymptomatic individuals whose offspring are at risk of a child with thalassemia major/ intermedia. The objective is to allow parents to make

reproductive preferences based on this information.

Who should be offered screening for thalassemia? This should be universal

### Antenatal Screening for Hemoglobinopathies<sup>8</sup>

A baseline screening should involve CBC and peripheral smear. Decreased red cell indices [mean corpuscular volume (MCV) < 80 fl and mean cell Hb (MCH) < 27 pg] with raised RBC counts suggests thalassemia trait. The screening test recommended for detection of carrier state of beta-thalassemia and structural Hemoglobin variants is high-performance liquid chromatography (HPLC) in cases with microcytosis. Decreased red cell indices [mean corpuscular volume (MCV) < 80 fl and mean cell Hb (MCH) < 27 pg] in association with HbA2  $\geq$  3.5 percent will detect most of the beta-thalassemia carriers and will also detect other Hb variants such as HbS, HbC, and HbE. These have clinically significant interactions with beta-thalassemia.

Women who are carrier with the Hb variants/ Thalassemia trait, their partner should be screened. When both partners are carriers of  $\beta$ -thalassemia/ Hb variants they should be referred for genetic counselling so as to decide prenatal diagnosis.

## Spinal Muscular Atrophy

Spinal muscular atrophy is an autosomal recessive disease characterized by degeneration of spinal cord motor neurons leading to atrophy of skeletal muscle and overall weakness, respiratory failure and death. The disorder is caused by a mutation in the gene known as the survival motor neuron gene (SMN1).

The incidence of spinal muscular atrophy is approximately 1 in 6,000 to 1 in 10,000 live births. Carrier frequencies in most populations are estimated at 1 in 40 to 1 in 60.

Who should be offered screening?

Screening for spinal muscular atrophy should be offered to all women who are considering pregnancy or are currently pregnant.

Spinal Muscular Atrophy carrier testing is done by MLPA.

## Fragile X Syndrome

Fragile X syndrome is the most common inherited form of mental retardation. Prevalence is approximately 1 in 3,600 males and 1 in 4,000-6,000 females from a variety of ethnic backgrounds.

Fragile X syndrome is transmitted as an X-linked disorder. The disorder is caused by expansion of a triple repeat CGG that leads to altered transcription of the fragile X gene FMR1.

### Who should be offered screening?

Fragile X premutation carrier screening is recommended

1. For women with a family history of fragile X-related disorders or intellectual disability.
2. If a woman has unexplained ovarian insufficiency or failure or an elevated follicle-stimulating hormone level before age 40 years

## B. EXPANDED CARRIER SCREENING<sup>9</sup>:

Significant advances in technology has led to simultaneous testing of large number of genes in one test. Expanded carrier screening refers to simultaneously screening for a large number of conditions. The majority of conditions are autosomal-recessive, but some may be X-linked or autosomal-dominant single gene disorders.

**TECHNIQUES:** The two primary laboratory techniques for testing are

- (1) Targeted high-throughput mutation analysis for specific mutations known to be associated with a particular disease
- (2) Sequence analysis via next-generation methodologies.

Screening for hundreds of disorders in one test could provide a potentially cost-effective means of identifying carriers for rare Mendelian disorders in the general population

The selection of disorders in panel is generally based on gene frequency and inclusion of mutations within a disorder that contribute to the highest detection of carriers.

Pre- and post-test counseling is essential to the process of carrier testing by a clinician trained in medical genetics.

Fig 1 shows approach for carrier screening disorder in the preconception/early pregnancy.

## Conclusions

Information on carrier evaluation should be provided to every pregnant woman. Ideally, screening and counseling are performed preconception, as this allows couples to

learn about their reproductive risk and reproductive options. Expanded carrier screening or screening for common genetic conditions - either of these should be offered. If it turns out that both partners are carriers of a genetic condition, genetic counseling should be offered.

## References

1. Baird PA, Anderson TW, Newcombe HB, Lowry RB. Genetic disorders in children and young adults: a population study. *Am J Hum Genet* 1988;42:677-93.
2. Verma IC, Puri RD, Global burden of genetic disease and the role of genetic screening, *Seminars in Fetal & Neonatal Medicine* (2015), <http://dx.doi.org/10.1016/j.siny.2015.07.002>
3. The Burden of Genetic Disorders in India and a Framework for Community Control I.C. Verma S. Bijarnia. *Community Genet* 2002;5:192-196
4. Ram KT, Klugman SD. Best practices: antenatal screening for common genetic conditions other than aneuploidy. *Curr Opin Obstet Gynecol* 2010;22(2):139-45.
5. The American College of Obstetricians and Gynecologists COMMITTEE OPINION Number 690 • March 2017 (Reaffirmed 2019)
6. Madan N, Sharma S, Sood SK, Colah R, Bhatia HM. The frequency of  $\beta$ -thalassemia trait and other hemoglobinopathies in northern and western India. *Indian J Hum Genet*. 2010;16:16-25.
7. Mohanty D, Colah RB, Gorakshakar AC, Patel RZ, Master DC, Mahanta J, et al. Prevalence of  $\beta$ -thalassemia and other haemoglobinopathies in six cities in India: A multicentre study. *J Community Genet*. 2013;4:33-42.
8. EMQN Best Practice Guidelines for molecular and haematology methods for carrier identification and prenatal diagnosis of the haemoglobinopathies. Joanne Traeger-Synodinos, Cornelis L Harteveld, John M Old, Mary Petrou, Renzo Galanello, Piero Giordano, Michael Angastioniotis, Barbara De la Salle, Shirley Henderson and Alison May on behalf of contributors to the EMQN haemoglobinopathies best practice meeting *European Journal of Human Genetics* (2015) 23, 426-437
9. Expanded carrier screening in pregnant women and women planning pregnancy :Britton D Rink, MD, MS, Louise Wilkins-Haug, MD, PhD, Vanessa A Barss, MD, FACOG. Jan 2019

## Forthcoming Events

- Next Monthly Clinical Meeting on 26<sup>th</sup> April, 2019 (4:00pm - 5:00pm) at Apollo Hospital.
- "Symposium on Chronic Pelvic Pain a Gynecologist's Perspective" at Max Hospital West Wing on 11<sup>th</sup> May 2019 (1:00pm - 3:00pm) at Max Saket West Wing auditorium under aegis of Multidisciplinary Committee of AOGD and DGFS

# Next Generation Sequencing in Obstetrics

Venkatesh Babu G, Madhulika Kabra, Neerja Gupta

Department of Pediatrics, Genetic Unit, Old OT Block, All India Institute of Medical Sciences, New Delhi



Dr Neerja Gupta

Genetic screening is necessary for clinical triage of a fetus for any birth defect. Traditionally for several decades it has involved maternal blood serum screening and ultrasound for soft markers to identify the women at risk for fetal aneuploidies and other structural birth defects. However, over recent years, rapid progress of genomics has revolutionized the field of prenatal diagnosis and fetal medicine. Next generation sequencing technologies are increasingly being used from screening till diagnosis to identify the underlying aetiology for an anomalous fetus.

## Next Generation Sequencing (NGS)

Human genome consists of about 3 billion base pairs and only ~1% of this entire genome is protein coding. The genome comprises of entire DNA content in both the coding (exonic) and non-coding (intronic) region. Exome represents the part of the genome which is formed by coding region transcribed and translated into proteins. Next generation sequencing involves massive parallel sequencing of the entire protein coding regions of about 20000 genes [whole exome sequencing (WES)] or sequencing of both exonic, intronic regions, non-coding RNA and mitochondrial DNA in addition to assessment of copy number variation and structural rearrangements known as whole genome sequencing (WGS) (van den Veyver IB, 2015). It is referred to as clinical exome sequencing, when it involves sequencing of coding regions of 4000-5000 genes known to be associated with human genetic disorders. Recent advances in next-generation sequencing (NGS) technologies, revealed significant decline in sequencing cost. This allows whole exome (WES) or targeted panels to be sequenced faster, at great depth and increased sensitivity, and this has made clinical application of WES more feasible. WES showed a diagnostic yield of approximately 30% when applied in routine clinical practice in adults and paediatric patients with structural anomaly and developmental delay (Carss et al., 2014).

### *The process of Next generation sequencing*

Beginning with the DNA extraction from whole blood, DNA is fragmented and subjected to library preparation. To 'capture' only expressed or protein coding region for exome sequencing, an enrichment procedure is required. This step is also used for capturing targeted gene panels allowing sequencing of only preselected genes in the panel.

### *Variant classification*

These techniques are high throughput and generate extensive data that requires careful interpretation

with respect to the clinical presentation. An inherently uneven yield of sequencing reads is observed across the genome and exome. Usually, an adequate coverage of approximately 85-98% is provided by an NGS technology for targeted sequence regions. Given the uncertainties of NGS results, the interpretation of the numerous variants arising as a result of NGS poses the biggest challenge. The American College of Medical Genetics and Genomics (ACMG) (Richards S, 2015) provides guidelines for accurate variant interpretation. Figure 2 (lower panel) illustrates the basic concept of whole exome and whole genome sequencing and five categories that are used for the interpretation of NGS results. These categories are pathogenic, likely pathogenic, variant of unknown significance, likely benign and benign.

- **Pathogenic-** A variant is identified to be disease causing. This result can be used to establish the definitive diagnosis.
- **Likely pathogenic-** A variant is identified to be significant based on the sufficient evidences with >90% certainty but there is no conclusive evidence to classify it as pathogenic.
- **Variant of unknown significance (VOUS)-** A variant is identified but no sufficient evidence to classify the variant as pathogenic or benign. Limited or conflicting evidence regarding the pathogenic effect of identified variant. Reassessment and further testing such as genetic, non-genetic clinical investigations or co-segregation analysis may be required to reclassify these variants as 'likely pathogenic' or 'likely benign'.
- **Likely benign-** A variant is identified to be insignificant based on the sufficient evidences but there is no conclusive evidence to classify it as benign. These are routinely not included in the clinical reports.
- **Benign-** A variant is not associated with disease based on sufficient evidences against its pathogenicity. These are routinely not included in the clinical reports.

**Validation-** The findings of exome sequencing (ES) are frequently validated by using Sanger sequencing or other technique depending upon the type of abnormality.

**Turnaround time -** The turnaround time in prenatal setting varies from 2-3 weeks (Best S, 2018) 2018. However, in our experience the turnaround time is not less than 4 weeks.

**Cost-** One of the main drawback is the exorbitant cost of the ES and it varies depending upon the type of exome (Whole vs Clinical) and across various companies.

A typical process for the next generation sequencing technique is beyond the scope of this article, however

a schematic diagram when a pregnant lady seeks genetic counselling is shown in figure 1. Pre and post-test counselling is an important and integral part of the management in this process. It involves providing information about the test, its potential results, turnaround time, need for further testing, the diagnostic yield and a followup testing. Couple should be clearly explained about the possibility of finding variant/s that could explain the fetal phenotype, possibility of getting a normal result meaning that molecular diagnosis is still not available for the fetal phenotype and the possibility of receiving an incidental finding that is not be related to the phenotype in question

## Utility of Next generation sequencing in obstetric practice

1. Noninvasive prenatal screening (NIPS) - Use of massively parallel technique in screening for aneuploidy from the cell free fetal DNA (cffDNA) from the maternal blood is known as Non-invasive prenatal screening (Palomaki GE 2011) (Bianchi DW 2012) (Vora NL 2018). It has been integrated into the clinical practice for about a decade and has revolutionized the approach to prenatal diagnosis. It is used as a screening test for the pregnant ladies with advanced maternal age, positive biochemical test for aneuploidies. It is most frequently used to detect aneuploidy in chromosomes 13, 18, 21, and the sex chromosomes either through a quantitative "counting" method that uses targeted parallel sequencing, or biallelic distributions ratio using single-nucleotide polymorphisms (SNPs). The allelic distribution method identifies the differences between maternal and fetal allele ratio. Equal proportion of each allele on a 1:1 ratio will be obtained for an heterozygous euploid fetus. On the contrary, a triploid fetus yields an allelic ratio of either 1:2 or 2:1 with a 90% sensitivity and 96% specificity. In Massively parallel sequencing, the entire genome will be sequenced by using NGS sequencing platform. The generated millions of short reads are reassembled against the human reference genome by using computational pipeline and each sequence of chromosomal origin is identified. The

percentage of reads are calculated by aligning to its specific chromosome and that is compared against the normal reference data to rule out that a chromosome is over or under represented (Figure 2 Upper panel). This method had specificity and sensitivity of 98.9% and 100.0%. Although frequently used for common aneuploidies, recently NIPS has been expanded to detect sub-chromosomal copy number variations (CNVs), of size lesser than 5MB such as involved in 16p12.2-p11.2, 1q21.1, 15q11.2, syndrome and 22q11 syndrome that are identical in unrelated individuals with conserved breakpoints. However, American College of Obstetrics and Gynaecology (ACOG) and the Society for Maternal Fetal Medicine (SMFM) do not recommend NIPS for microdeletion and microduplication syndromes due to high false positive and negative rates. The sensitivity of detection of the CNVs are affected by three independent factors and are as follows;

- Fetal fraction- Higher the fetal fraction, easier to detect.
  - Size of the CNV-Larger CNVs are detected easily. Recently, several different algorithms are available for detecting small CNVs.
  - Depth of coverage- As the coverage increases, CNVs of smaller sizes are detected.
2. Prenatal whole exome sequencing for fetal structural malformations- Identification of any fetal anomaly on antenatal ultrasound prompts one to classify the malformation either as isolated or multiple to better classify it etiologically and plan a genomic testing strategy (chromosomal analysis or chromosomal microarray, or a targeted gene or exome analysis) to offer proper counselling. WES is widely used in clinics postnatally, however, it is now being performed in selected cases on a research basis or, less commonly, on a clinical basis, where standard genetic testing fails to reveal a diagnosis. A recent systematic review of Exome sequencing in fetuses with structural malformation by Best et al (Best S 2018) revealed a broad range (6.2- 80%) of diagnostic yield across the 16 studies with five or more fetuses tested. A large UK based prospective cohort study( PAGE Prenatal

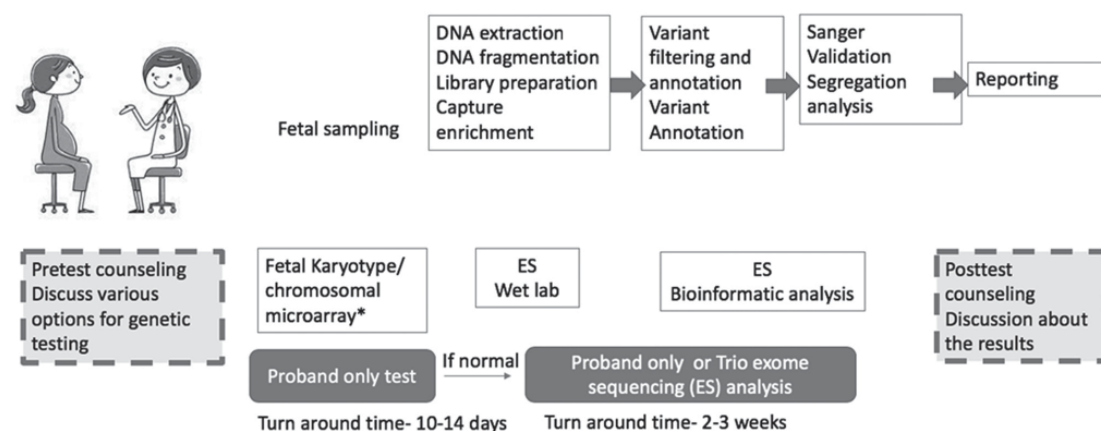


Figure 1 Process of genomic testing when a pregnant lady carrying fetus with birth defect with a probable underlying genetic etiology seeks a genetic referral

Assessment of Genome and Exome) of 610 fetuses with structural anomalies and their parents (parent-fetus trios) evaluated exome sequencing in assessing the pathogenic genetic variants in developmental disorder genes (the Deciphering Developmental Disorders study) (Wright CF 2018) after excluding chromosomal aneuploidies and large copy number variations showed an additional diagnostic yield of 8.5% when compared to conventional genetic testing (Lord et al., 2019). Another USA based prospective cohort study by Petrovski et al (Petrovski et al., 2019) evaluated 231 fetuses with structural anomalies using trio WES and achieved a diagnostic yield of 10.3%. Both studies differ as the former used a virtual panel of 1628 genes and the latter included all genes. In a prenatal setting, the diagnostic yield of WES in large prospective cohort studies with broad range of fetal structural anomalies was found to be less than the smaller retrospective studies on selected groups. Foetuses with multisystem anomalies showed more pathogenic/likely pathogenic variants than compared to isolated anomalies (Jelin and Vora, 2018).

Recent guidance from the American College of Obstetricians and Gynaecologists and a Joint Position Statement on the use of genome- wide sequencing for fetal diagnosis *does not* recommend the routine use of exome sequencing in the prenatal setting as a diagnostic test. It further suggests that it may be considered on case to case basis with expert guidance from the geneticists. It is better to perform chromosomal microarray prior to the whole exome sequencing for fetal structural anomalies. In fetal abnormalities specific for a genetic syndrome, a targeted genetic testing should be performed. The indications of prenatal exome sequencing are shown in box 1.

**Box 1: Indications for prenatal exome (Diagnosis, 2018, Jelin and Vora, 2018, Vora and Hui, 2018)**

1. Recurrent single or multiple congenital anomalies in the fetus with a possible monogenic etiology with normal karyotype and/or chromosomal microarray in the anomalous fetus
2. Undiagnosed skeletal dysplasia
3. Multiple congenital anomalies in a fetus with history of recurrent stillbirths with normal karyotype and/or chromosomal microarray
4. Fetus with structural abnormalities with reported consanguinity or homozygosity reported on CMA.
5. Known structural abnormalities associated with high genetic heterogeneity (Vora and Hui, 2018).

**Fetal Exome and Gene discovery-** The utilisation of WES in prenatal screening is increasing the interest of identifying novel candidate genes critical to human development. By using prenatal WES in a family with two sibs affected by a perinatal lethal short rib-polydactyly syndrome III, compound heterozygous mutations were identified in *WDR60* which is a relatively uncharacterized gene. Compound heterozygous mutations of *WDR60*

were confirmed in another unrelated Spanish individual with Jeune syndrome after analysis of an additional 54 skeletal ciliopathy exomes. Further functional assays confirmed, *WDR60* mutations can cause skeletal ciliopathies and suggest a role for *WDR60* in ciliogenesis (McInerney-Leo et al., 2013).

In a study of Caucasian origin, two sibling reported with undescribed lethal fetal congenital anomaly syndrome which included intrauterine growth restriction, severe microcephaly, renal cystic dysplasia/agenesis, and complex brain and genitourinary malformations with non-consanguineous family background suggesting a possible ciliopathy, but was not indicative of any known condition. Novel autosomal recessive truncating mutations in *KIF14* gene was identified on a family based study using whole exome sequencing that segregated with the phenotype. Several mice model studies also demonstrated autosomal recessive mutations in the *KIF14* (Filges et al., 2014) expressing unusually a similar phenotype (Fujikura et al., 2013).

Three female infants from a non-consanguineous family died with an extreme form of microcephaly with apparent cerebral growth arrest between 14 and 18 weeks' gestation were screened by WES identified

**Box 2: Advantages and limitations of WES**

*Advantages*

1. Ends diagnostic odyssey and additional diagnostic yield
2. Identification of new genes and phenotypes

*Limitations*

1. Time compressed situation with multiple sample processing such as prior maternal cell contamination, bioinformatic analysis and sanger validation
2. DNA quality and quantity
3. Incomplete exon capture and problems of GC rich regions
4. Limited phenotypic information relying mainly on the sonologist findings
5. Interpretation related challenges such as variant of unknown significance, incidental findings, false negative results
6. Genotype phenotype correlation\*

*Not useful for the detection of*

1. Chromosomal abnormalities
2. Triplet repeat disorders
3. Imprinting disorders
4. Large deletions and duplications

\*The major challenge for prenatal WES is genotype phenotype correlation. As the information about the fetal phenotype is limited to the ultrasonographic, fetal ECHO or fetal MRI findings, the genotype phenotype correlation may not be accurate. A postnatal evaluation of the baby or fetal autopsy in case of termination is usually necessary to correlate with the genotype. Postnatal databases such as ClinGen and Matchmaker like databases are required to examine similar phenotypes thereby increasing the confidence in pursuing novel gene discovery functional studies in multiple families with genotype/phenotype correlations. At present there are no such available shared databases for defects of prenatal origin.

homozygous mutations in *MKL2*. Screening of another 51 cases with varying degree of microcephaly could not identify variants in this gene (Ramos et al., 2014). Functional studies confirmed disruption of *MKL2*:SRF axis produced severe microcephaly. Putoux et al. by using both combination of homozygosity mapping and targeted sequencing of four affected fetuses with hydroletharus and acrocallosal syndromes and two multiple malformation disorders with overlapping features such as cleft palate, polydactyly, brain abnormalities identified mutations in *KIF7* pooled with identifying a truncating variant of the same gene in eight individuals (Putoux et al., 2011). Another study reported a similar approach to identify *TCTN3* as the cause of orofacioidigital (OFD) syndrome IV (Mohr-Majewski syndrome) (Thomas et al., 2012).

Box 2 enlists the advantages and limitations of WES.

### Prenatal whole Genome Sequencing (WGS)

Whole genome sequencing has been applied in selected prenatal cases but has not been implicated clinically because of the need to pay more attention to analysis of massive amount of information (intronic/regulatory regions) it will deliver to parents (Talkowski et al., 2012). WGS overcome the limitation of WES like coding exon coverage insufficiency (e.g., GC-rich exons) by polymerase chain reaction (PCR)-free technology

(Meienberg et al., 2016, Belkadi et al., 2015). WGS has additional great ability to detect copy-number variants (CNVs), (Gilissen et al., 2014, Handsaker et al., 2015) and other structural variants. WGS also provides higher sensitivity to express of short tandem repeats than WES (Dolzhenko et al., 2017). Prenatal whole genome sequencing is gaining attention as a diagnostic and discovery strategy in suspected genetic disorder that remains undiagnosed after WES. Recent WGS study identified additional causative variants in both coding and noncoding regions in autism disorder and recognized 3% more protein coding variants which are missed by WES (Turner et al., 2017). Mao et al performed a comparative WGS study on 31 fetuses on amniotic fluid isolated cfDNA and cell pellet isolated DNA. Compare to cell pellet isolated DNA, cfDNA also able to generate the data to find potentially detrimental variants. (Mao et al., 2018)

Figure 2 shows a diagrammatic representation of NIPS and NGS. Upper panel represents the NGS based approach for NIPS of fetal aneuploidy where X-axis is showing the number of targets from each chromosome (coloured dots) and Y-axis is showing z-score of each chromosome (Ch21 showing higher proportion). Lower panel represents the NGS based whole exome and whole genome sequencing approach and variant classification as per ACMG guidelines.

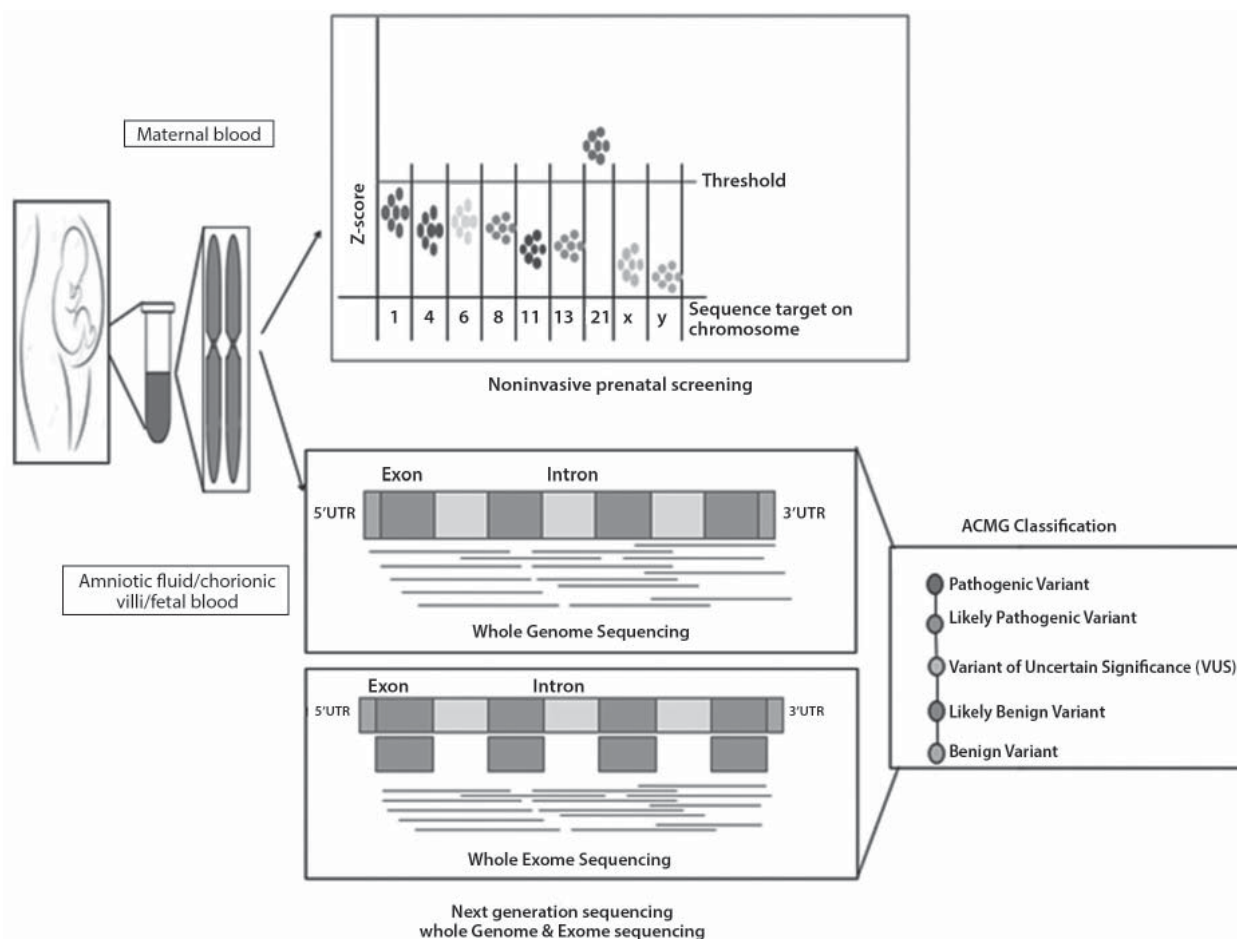


Figure 2

## Important points to remember while ordering NGS in a prenatal setting

- Pre and posttest counselling about the diagnostic yield, cost, implications of test results and possibility of a normal report or detection of an incidental medically relevant finding for a disorder not in question.
- Implications of variant of unknown significance that may or may not be related to the fetal phenotype and might need the follow up evaluation.
- Validation of the positive findings by reverse phenotyping and Sanger sequencing.

## Conclusions

In conclusion, NGS is a valuable and powerful tool in ending the diagnostic odyssey in the field of medicine. Obstetricians need to be well versed with the basic principles of next generation sequencing, as it would be one of the most frequently used technique in the prenatal setting in next few years in the developing countries. Diagnostic rates, test cost and turnaround time varies based upon the type of exome. Accurate phenotyping, correct choice of test and its interpretation is vital for establishing a fetal diagnosis. Detailed pretest and posttest counselling is crucial while offering NGS.

## References

- Alamillo, C L, Powis, Z, Farwell, K, Shahmirzadi, L, Weltmer, E C, Turocy, J, Lowe, T, Kobelka, C, Chen, E, Basel, D, Ashkinadze, E, D'augelli, L, Chao, E & Tang, S 2015. Exome sequencing positively identified relevant alterations in more than half of cases with an indication of prenatal ultrasound anomalies. *Prenat Diagn*, 35, 1073-8.
- Belkadi, A, Bolze, A, Itan, Y, Cobat, A, Vincent, Q B, Antipenko, A, Shang, L, Boisson, B, Casanova, J L & Abel, L 2015. Whole-genome sequencing is more powerful than whole-exome sequencing for detecting exome variants. *Proc Natl Acad Sci U S A*, 112, 5473-8.
- Bl, W, Breman, A, Shaw, C A, Stankiewicz, P, Gambin, T, Lu, X, Cheung, S W, Jackson, L G, Lupski, J R, Van Den Veyver, I B & Beaudet, A L 2012. Detection of  $\geq 1$ Mb microdeletions and microduplications in a single cell using custom oligonucleotide arrays. *Prenat Diagn*, 32, 10-20.
- Breman, A M, Chow, J C, U'ren, L, Normand, E A, Qdaisat, S, Zhao, L, Henke, D M, Chen, R, Shaw, C A, Jackson, L, Yang, Y, Vossaert, L, Needham, R H, Chang, E J, Campton, D, Werbin, J L, Seubert, R C, Van Den Veyver, I B, Stilwell, J L, Kaldjian, E P & Beaudet, A L 2016. Evidence for feasibility of fetal trophoblastic cell-based noninvasive prenatal testing. *Prenat Diagn*, 36, 1009-1019.
- Carss, K J, Hillman, S C, Parthiban, V, McMullan, D J, Maher, E R, Kilby, M D & Hurler, M E 2014. Exome sequencing improves genetic diagnosis of structural fetal abnormalities revealed by ultrasound. *Hum Mol Genet*, 23, 3269-77.
- Dolzhenko, E, Van Vugt, J, Shaw, R J, Bekritsky, M A, Van Blitterswijk, M, Narzisi, G, Ajay, S S, Rajan, V, Lajoie, B R, Johnson, N H, Kingsbury, Z, Humphray, S J, Schellevis, R D, Brands, W J, Baker, M, Rademakers, R, Kooyman, M, Tazelaar, G H P, Van Es, M A, McLaughlin, R, Sproviero, W, Shatunov, A, Jones, A, Al Khleifat, A, Pittman, A, Morgan, S, Hardiman, O, Al-Chalabi, A, Shaw, C, Smith, B, Neo, E J, Morrison, K, Shaw, P J, Reeves, C, Winterkorn, L, Wexler, N S, Housman, D E, Ng, C W, Li, A L, Taft, R J, Van Den Berg, L H, Bentley, D R, Veldink, J H & Eberle, M A 2017. Detection of long repeat expansions from PCR-free whole-genome sequence data. *Genome Res*, 27, 1895-1903.
- Diagnosis, ISFP 2018. Joint Position Statement from the International Society for Prenatal Diagnosis (ISPD), the Society for Maternal Fetal Medicine (SMFM), and the Perinatal Quality Foundation (PQF) on the use of genome-wide sequencing for fetal diagnosis. *Prenat Diagn*, 38, 6-9.
- Drury, S, Williams, H, Trump, N, Boustred, C, Lench, N, Scott, R H & Chitty, L S 2015. Exome sequencing for prenatal diagnosis of fetuses with sonographic abnormalities. *Prenat Diagn*, 35, 1010-7.
- Fan, H C, Gu, W, Wang, J, Blumenfeld, Y J, El-Sayed, Y Y & Quake, S R 2012. Non-invasive prenatal measurement of the fetal genome. *Nature*, 487, 320-4.
- Filges, I, Nosova, E, Bruder, E, Tercanli, S, Townsend, K, Gibson, W T, Rothlisberger, B, Heinemann, K, Hall, J G, Gregory-Evans, C Y, Wasserman, W W, Miny, P & Friedman, J M 2014. Exome sequencing identifies mutations in KIF14 as a novel cause of an autosomal recessive lethal fetal ciliopathy phenotype. *Clin Genet*, 86, 220-8.
- Fiorentino, F, Napoletano, S, Caiazzo, F, Sessa, M, Bono, S, Spizzichino, L, Gordon, A, Nuccitelli, A, Rizzo, G & Baldi, M 2013. Chromosomal microarray analysis as a first-line test in pregnancies with a priori low risk for the detection of submicroscopic chromosomal abnormalities. *Eur J Hum Genet*, 21, 725-30.
- Fujikura, K, Setsu, T, Tanigaki, K, Abe, T, Kiyonari, H, Terashima, T & Sakisaka, T 2013. Kif14 mutation causes severe brain malformation and hypomyelination. *PLoS One*, 8, e53490.
- Gilissen, C, Hehir-Kwa, J Y, Thung, D T, Van De Vorst, M, Van Bon, B W, Willemsen, M H, Kwint, M, Janssen, I M, Hoischen, A, Schenck, A, Leach, R, Klein, R, Tearle, R, Bo, T, Pfundt, R, Yntema, H G, De Vries, B B, Kleefstra, T, Brunner, H G, Vissers, L E & Veltman, J A 2014. Genome sequencing identifies major causes of severe intellectual disability. *Nature*, 511, 344-7.
- Handsaker, R E, Van Doren, V, Berman, J R, Genovese, G, Kashin, S, Boettger, L M & McCarroll, S A 2015. Large multiallelic copy number variations in humans. *Nat Genet*, 47, 296-303.
- Jelin, A C & Vora, N 2018 Whole Exome Sequencing: Applications in Prenatal Genetics. *Obstet Gynecol Clin North Am*, 45, 69-81.
- Kitzman, J O, Snyder, M W, Ventura, M, Lewis, A P, Qiu, R, Simmons, L E, Gammill, H S, Rubens, C E, Santillan, D A, Murray, J C, Tabor, H K, Bamshad, M J, Eichler, E E & Shendure, J 2012. Noninvasive whole-genome sequencing of a human fetus. *Sci Transl Med*, 4, 137ra76.
- Lord, J, McMullan, D J, Eberhardt, R Y, Rinck, G, Hamilton, S J, Quinlan-Jones, E, Prigmore, E, Keelagher, R, Best, S K, Carey, G K, Mellis, R, Robart, S, Berry, I R, Chandler, K E, Cilliers, D, Cresswell, L, Edwards, S L, Gardiner, C, Henderson, A, Holden, S T, Homfray, T, Lester, T, Lewis, R A, Newbury-Ecob, R, Prescott, K, Quarrell, O W, Ramsden, S C, Roberts, E, Tapon, D, Tooley, M J, Vasudevan, P C, Weber, A P, Wellesley, D G, Westwood, P, White, H, Parker, M, Williams, D, Jenkins, L, Scott, R H, Kilby, M D, Chitty, L S, Hurler, M E & Maher, E R 2019. Prenatal exome sequencing analysis in fetal structural anomalies detected by ultrasonography (PAGE): a cohort study. *Lancet*, 393, 747-757.

- Mao, Q, Chin, R, Xie, W, Deng, Y, Zhang, W, Xu, H, Zhang, R Y, Shi, Q, Peters, E E, Gulbahce, N, Li, Z, Chen, F, Drmanac, R & Peters, B A 2018. Advanced Whole-Genome Sequencing and Analysis of Fetal Genomes from Amniotic Fluid. *Clin Chem*, 64, 715-725.
- Mcinerney-Leo, A M, Schmidts, M, Cortes, C R, Leo, P J, Gener, B, Courtney, A D, Gardiner, B, Harris, J A, Lu, Y, Marshall, M, Scambler, P J, Beales, P L, Brown, M A, Zankl, A, Mitchison, H M, Duncan, E L & Wicking, C 2013. Short-rib polydactyly and Jeune syndromes are caused by mutations in WDR60. *Am J Hum Genet*, 93, 515-23.
- Meienberg, J, Bruggmann, R, Oexle, K & Matyas, G 2016. Clinical sequencing: is WGS the better WES? *Hum Genet*, 135, 359-62.
- Petrovski, S, Aggarwal, V, Giordano, J L, Stosic, M, Wou, K, Bier, L, Spiegel, E, Brennan, K, Stong, N, Jobanputra, V, Ren, Z, Zhu, X, Mebane, C, Nahum, O, Wang, Q, Kamalakaran, S, Malone, C, Anyane-Yeboah, K, Miller, R, Levy, B, Goldstein, D B & Wapner, R J 2019. Whole-exome sequencing in the evaluation of fetal structural anomalies: a prospective cohort study. *Lancet*, 393, 758-767.
- Putoux, A, Thomas, S, Coene, K L, Davis, E E, Alanay, Y, Ogur, G, Uz, E, Buzas, D, Gomes, C, Patrier, S, Bennett, C L, Elkhartoufi, N, Frison, M H, Rigonnot, L, Joye, N, Pruvost, S, Utine, G E, Boduroglu, K, Nitschke, P, Fertitta, L, Thauvin-Robinet, C, Munnich, A, Cormier-Daire, V, Hennekam, R, Colin, E, Akarsu, N A, Bole-Feysot, C, Cagnard, N, Schmitt, A, Goudin, N, Lyonnet, S, Encha-Razavi, F, Siffroi, J P, Winey, M, Katsanis, N, Gonzales, M, Vekemans, M, Beales, P L & Attie-Bitach, T 2011. KIF7 mutations cause fetal hydrolethrus and acrocallosal syndromes. *Nat Genet*, 43, 601-6.
- Ramos, E I, Bien-Willner, G A, Li, J, Hughes, A E, Giacalone, J, Chasnoff, S, Kulkarni, S, Parmacek, M, Cole, F S & Druley, T E 2014. Genetic variation in MKL2 and decreased downstream PCTAIRE1 expression in extreme, fatal primary human microcephaly. *Clin Genet*, 85, 423-32.
- Talkowski, M E, Ordulu, Z, Pillalamarri, V, Benson, C B, Blumenthal, I, Connolly, S, Hanscom, C, Hussain, N, Pereira, S, Picker, J, Rosenfeld, J A, Shaffer, L G, Wilkins-Haug, L E, Gusella, J F & Morton, C C 2012. Clinical diagnosis by whole-genome sequencing of a prenatal sample. *N Engl J Med*, 367, 2226-32.
- Thomas, S, Legendre, M, Saunier, S, Bessieres, B, Alby, C, Bonniere, M, Toutain, A, Loeuillet, L, Szymanska, K, Jossic, F, Gaillard, D, Yacoubi, M T, Mougou-Zerelli, S, David, A, Barthez, M A, Ville, Y, Bole-Feysot, C, Nitschke, P, Lyonnet, S, Munnich, A, Johnson, C A, Encha-Razavi, F, Cormier-Daire, V, Thauvin-Robinet, C, Vekemans, M & Attie-Bitach, T 2012. TCTN3 mutations cause Mohr-Majewski syndrome. *Am J Hum Genet*, 91, 372-8.
- Turner, T N, Coe, B P, Dickel, D E, Hoekzema, K, Nelson, B J, Zody, M C, Kronenberg, Z N, Hormozdiari, F, Raja, A, Pennacchio, L A, Darnell, R. B & Eichler, E E 2017. Genomic Patterns of De Novo Mutation in Simplex Autism. *Cell*, 171, 710-722 e12.
- Vora, N L & HUI, L 2018. Next-generation sequencing and prenatal 'omics: advanced diagnostics and new insights into human development. *Genet Med*, 20, 791-799.
- Wapner R, P S, Brennan K, et al 2017;. 8: Whole exome sequencing in the evaluation of fetal structural anomalies: a prospective study of sequential patients. *Am J Obstet Gynecol*, 216:S5-S6.
- Wapner, R J, Martin, C. L, Levy, B, Ballif, B C, Eng, C M, Zachary, J M, Savage, M, Platt, L D, Saltzman, D, Grobman, W A, Klugman, S, Scholl, T, Simpson, J L, Mccall, K, Aggarwal, V S, Bunke, B, Nahum, O, Patel, A, Lamb, A N, Thom, E A, Beaudet, A L, Ledbetter, D H, Shaffer, L G & Jackson, L 2012. Chromosomal microarray versus karyotyping for prenatal diagnosis. *N Engl J Med*, 367, 2175-84.

## CONTROVERSY

# Can Non-Invasive Prenatal Testing (NIPT) replace Biochemical Screening ?



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## Introduction

Prenatal screening for has undergone rapid changes in the last few decades. Initially screening was based on second trimester biochemistry and then the focus shifted to first trimester biochemistry in the 1990s. Introduction of non-invasive prenatal screening (NIPS) using cell free DNA in maternal blood has been a paradigm shift in the screening armamentarium.

## Biochemical screening in the first trimester

### First trimester 'dual marker'

The two most extensively evaluated maternal serum markers in the first trimester are free beta human chorionic gonadotropin (beta hCG) and pregnancy associated plasma protein A (PAPP-A). hCG is glycoprotein formed by the developing embryo and later by the placenta. It can be assayed in both free and total forms. PAPP-A is a complex high molecular weight glycoprotein. Both can be measured between 9 weeks to 13 weeks and 6 days and are effective screening test for Down syndrome. The risk is generated by converting the maternal test result into MoMs (multiple of median) and by adjusting it for maternal age, maternal weight, gestational age, ethnicity, smoking, type of conception, diabetes, number of fetuses and chorionicity. This adjustment is essential for calculation of accurate risks.<sup>1</sup> The expected changes in these levels with the common aneuploidies is given in table 1.

Table 1: Expected biochemistry levels in euploid and aneuploid fetuses<sup>2,3</sup>

Study population	Median free beta hCG (MoM)	Median PAPP-A (MoM)
Euploid	1.0	1.0
Trisomy 21	2.0	0.5
Trisomy 18	0.2	0.2
Trisomy 13	0.5	0.3
Turner syndrome	1.0	0.4

MoM: multiple of median

When combined with ultrasound markers especially an accurately measured nuchal translucency (NT), the detection rate for Down syndrome is 85% for a false positive rate of 5%.<sup>4,5</sup> Addition of more ultrasound markers further improves the detection rate and brings down the false positive rate: the combination of maternal age-related risk, fetal NT, and serum markers yields a trisomy 21 detection rate of 90% for a false positive rate of 3-5%.<sup>6</sup> The detection rate in screening for

trisomy 18 and 13 is about 95%.<sup>6</sup> Effective performance of this screening strategy has been established in the North Indian population.<sup>7</sup>

### First trimester 'quadruple marker'

The addition of maternal serum markers, i.e. alphafetoprotein (AFP) and placental growth factor (PlGF) has also been shown to improve detection rates to 93% while bringing down the false positive rate to 1-1.5%.<sup>8,9</sup> Addition of PlGF is of particular importance as it has been proven to be an effective screening tool for development of early preeclampsia (necessitating delivery before 34 weeks).<sup>10</sup> This assumes greater significance in light of the fact that there is level I evidence that low dose aspirin (150 mg once a day) can prevent 80% of early preeclampsia in women who are screen positive using the FMF algorithm.<sup>11</sup>

### Second trimester triple and quadruple marker

Women who present late in the pregnancy and/or have missed their dual marker can be offered either a triple or a quadruple test up to 22 weeks. Triple marker includes serum total beta hCG, estriol and AFP and has the least detection rate of 60-65%. Quadruple marker adds inhibin A to the previous three hormones with an improved detection rate of 75-80%.<sup>4</sup> AFP in the second trimester may have the added advantage of picking up women with open neural tube defects; however the ultrasound detection rate for open neural tube defects is nearly 100%. It is important to note that the detection rate for Down syndrome on second trimester anomaly scan is a dismal 58% and hence addition of serum biochemistry is recommended despite the high false positive rates.<sup>12</sup>

Table 2: Detection rates and Odds of being affected given a positive result (OAPR) for various screening models for Down syndrome<sup>4,12,13,14</sup>

Method	Detection rate	OAPR
<b>Second trimester</b>		
Triple	69%	
Quadruple	75-80%	1 in 36
Anomaly scan	56%	1 in 41
Quadruple + Anomaly scan	80%	1 in 29
<b>First trimester</b>		
Combined	82-85%	1 in 29
Combined + NB	91%	1 in 26
Combined + PlGF and AFP	90%	1 in 44
<b>Both trimesters</b>		
Integrated	92%	1 in 42
Contingent	91%	1 in 42
<b>NIPT/NIPS</b>	<b>99.9</b>	<b>2 in 3</b>

## Noninvasive prenatal screening/test (NIPS/ NIPT)

NIPT or cell-free fetal DNA (cffDNA) is currently the best screening test available for prenatal detection of trisomy 21 in singleton pregnancy. The latest meta-analysis by Gil et al. reports a weighted pooled detection rate (DR) of 99.2 % and a false positive rate (FPR) of 0.09 %.<sup>14</sup> A recently published large, multicentric trial has reported higher sensitivity (100 % DR), lower FPR (0.06 %), and higher PPV (80.9 %) for cffDNA testing for trisomy 21 compared to standard screening with first trimester nuchal translucency and biochemistry in routine prenatal screening population.<sup>15</sup> Thus, cffDNA is indeed the best screening test for trisomy 21 even in low-risk population. An important factor in maintaining the high DR is fetal fraction (cut-off of 4 % for most labs). If the test cannot be reported due to low fetal fraction, the possibility of aneuploidy is higher and invasive testing viz a viz repeat sampling should be discussed. Fetal fraction is also likely to be low in women with high BMI and these women should be counseled accordingly. Fetal fraction is sufficient for reporting after 10 weeks' gestation. The turnaround time for cfDNA is 7-10 days; thus gestational age will be important in deciding for the test as one would like to have a result well before 20 weeks to allow time for invasive testing if needed. However, NIPT is still a 'screening test' and the decision to terminate must not be based on it. All high-risk/positive results must be offered invasive testing to confirm (or indeed rule out) aneuploidy. This should preferably be by amniocentesis to minimize errors due to placental mosaicism.<sup>16</sup>

### NIPT in place of biochemistry for all?

Although NIPT is currently the best screening test for Down syndrome in singleton pregnancies, it is unlikely

to replace serum biochemistry for two main reasons: its significant cost precludes its use as a primary screening modality in public health setting. The most economical way of implementing this costly test would be in a contingent manner: all women should be offered combined first trimester screening by ultrasound, PAPP-A, and beta hCG. Women with very high risk should be offered invasive testing, whereas women with intermediate risk should be offered NIPT. This policy would require NIPT for 1/4th of the screened population and detect 98% of trisomies at an invasive rate of only 0.8 %.<sup>17</sup> This view has also been endorsed by the AOGD Good Clinical Practice Recommendations on Aneuploidy Screening in Pregnancy drafted by the AOGD Fetal Medicine Subcommittee (2017 - 2019). The suggested flowcharts by the subcommittee for desirable screening protocols in Indian population are given in figures 1 and 2.

It is pertinent to highlight here that neither NIPT nor serum biochemistry should be a replacement for properly performed nuchal scan which not only provides better detection rates for aneuploidies but also detects structural abnormalities whose incidence is higher than the incidence of Down syndrome.<sup>19,20</sup>

## Conclusion

Non-invasive prenatal screening is undoubtedly the best screening test for Down syndrome; however, it is unlikely to replace first trimester serum biochemistry both because of the costs involved and also because it cannot provide risk assessment for early preeclampsia/ fetal growth restriction. The most effective way of utilizing this excellent albeit screening test would be in a contingent manner.

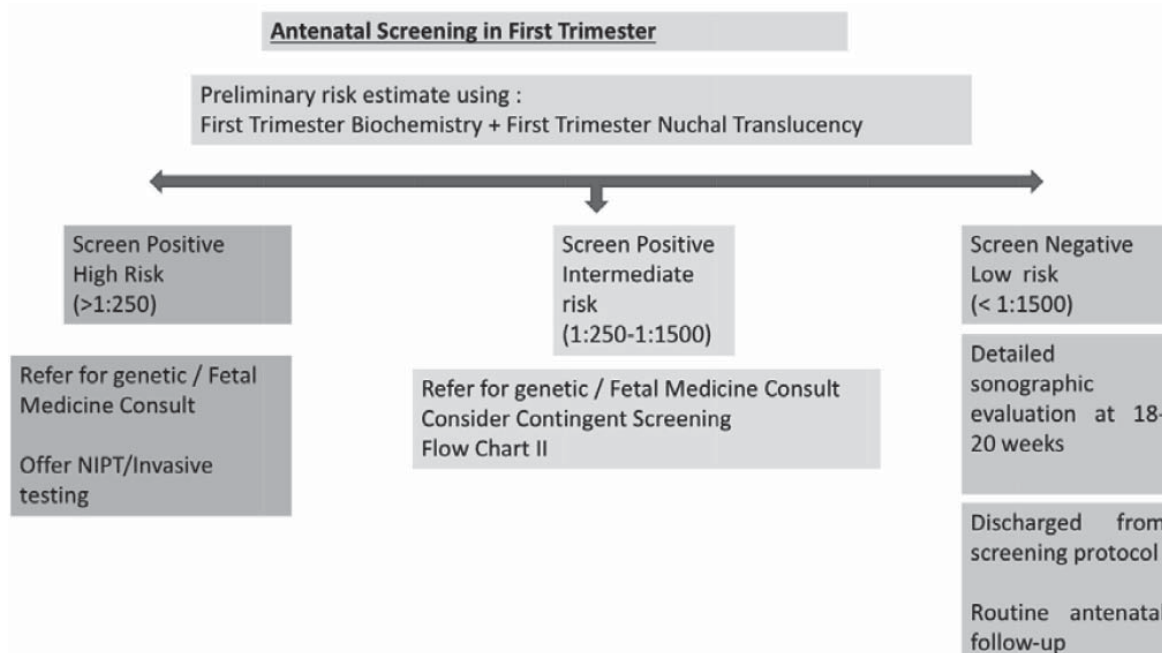


Figure 1: Desirable screening protocol in the first trimester

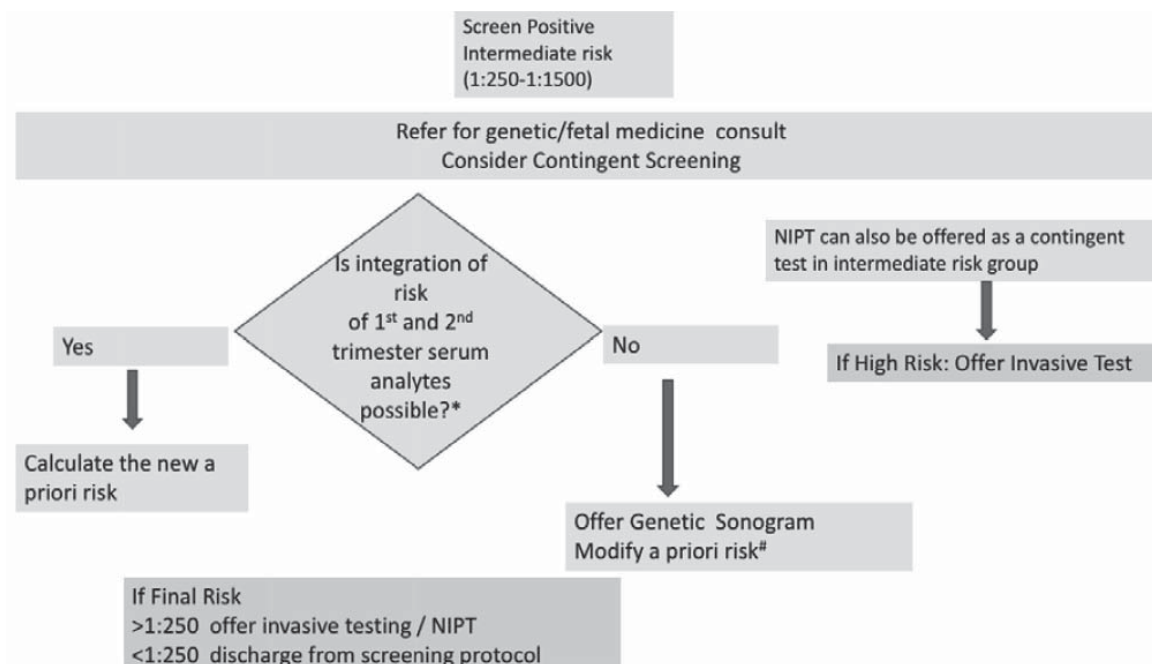


Figure 2: Contingent Screening in Intermediate Risk Group

## References

1. Kagan KO, Wright D, Spencer K et al. First-trimester screening for trisomy 21 by free beta-human chorionic gonadotropin and pregnancy-associated plasma protein-A: impact of maternal and pregnancy characteristics. *Ultrasound Obstet Gynecol* 2008; 31(5):493-502.
2. Wagner P, Sonek J, Hoopmann M et al. First-trimester screening for trisomies 18 and 13, triploidy and Turner syndrome by detailed early anomaly scan. *Ultrasound Obstet Gynecol* 2016 Oct; 48(4): 446-451.
3. Kagan KO, Sonek J, Wagner P, Hoopmann M. Principles of first trimester screening in the age of noninvasive prenatal diagnosis: screening for chromosomal abnormalities. *Arch Gynecol Obstet*. 2017 Oct; 296(4): 645-651.
4. Malone FD, Canick JA, Ball RH, et al. First- and Second-Trimester Evaluation of Risk (FASTER) Research Consortium. First-trimester or second-trimester screening, or both, for Down's syndrome. *N Engl J Med* 2005;353:2001e11.
5. Wald NJ, Rodeck C, Hackshaw AK, et al. First and second trimester antenatal screening for Down's syndrome: the results of the Serum, Urine and Ultrasound Screening Study (SURUSS). *J Med Screen* 2003;10:56e104.
6. Santorum M, Wright D, Syngelaki A et al (2016) Accuracy of first trimester combined test in screening for trisomies 21, 18 and 13. *Ultrasound Obstet Gynecol*.
7. Kaul A, Singh C, Gupta R. Observational study comparing the performance of first trimester screening protocols for detecting trisomy 21 in a North Indian population. *Int J Gynaecol Obstet*. 2017 Apr;137(1):14-19.
8. Kagan KO, Hoopmann M, Abele H et al (2012) First-trimester combined screening for trisomy 21 with different combinations of placental growth factor, free B-human chorionic gonadotropin and pregnancy-associated plasma protein-A. *Ultrasound Obstet Gynecol* 40:530-535.
9. Bredaki FE, Wright D, Matos P et al (2011) First-trimester screening for trisomy 21 using alpha-fetoprotein. *Fetal Diagn Ther* 30:215-218.
10. Pandya P, Wright D, Syngelaki A et al (2012) Maternal serum placental growth factor in prospective screening for aneuploidies at 8-13 weeks' gestation. *Fetal Diagn Ther* 31: 87-93.
11. Rolnik DL, Wright D, Poon LCY, et al. ASPRE trial: performance of screening for preterm pre-eclampsia. *Ultrasound Obstet Gynecol*. 2017 Oct;50(4):492-495.
12. Benn P, Borrell A, Chiu RW, et al. Position statement from the Chromosome Abnormality Screening Committee on behalf of the Board of the International Society for Prenatal Diagnosis. *Prenat Diagn*. 2015 Aug;35(8):725-34.
13. Cuckle H, Maymon R (2016) Development of prenatal screening—a historical overview. *Semin Perinatol* 40:12-22.
14. Gil MM, Quezada MS, Revello R, et al. Analysis of cell-free DNA in maternal blood in screening for fetal aneuploidies: updated meta-analysis. *Ultrasound Obstet Gynecol*. 2015; 45: 249-66.
15. Norton ME, Jacobsson B, Swamy GK, et al. Cell-free DNA analysis for noninvasive examination of trisomy. *N Engl J Med*. 2015;372(17):1589-97.
16. Everett TR, Chitty LS. Cell-free fetal DNA: the new tool in fetal medicine. *Ultrasound Obstet Gynecol*. 2015; 45(5): 499-507.
17. Committee opinion no 640: cell free DNA screening for aneuploidies. American College of Obstetricians and Gynecologists Committee on Genetics. *Obstet Gynecol*. 2015, Jun 29.
18. Gil MM, Akolekar R, Quezada MS, et al. Analysis of cell-free DNA in maternal blood in screening for aneuploidies: meta-analysis. *Fetal Diagn Ther*. 2014;35:156-73.
19. Alldred SK, Takwoingi Y, Guo B, et al. First trimester ultrasound tests alone or in combination with first trimester serum tests for Down's syndrome screening. *Cochrane Database Syst Rev*. 2017 Mar 15;3:CD012600.
20. Salomon LJ, Alfirevic Z, Audibert F, et al. ISUOG Clinical Standards Committee. ISUOG consensus statement on the impact of non-invasive prenatal testing (NIPT) on prenatal ultrasound practice. *Ultrasound Obstet Gynecol*. 2014 Jul;44(1):122-3.

## CASE APPROACH

# Nonimmune Hydrops Fetalis

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Dr Manisha Kumar

## What is non immune hydrops?

Hydrops fetalis is defined as the accumulation of abnormal fluid in at least two different fetal compartments. It generally presents as subcutaneous edema, accompanied by effusions in two or more serous cavities including pericardial or pleural effusions, and ascites.

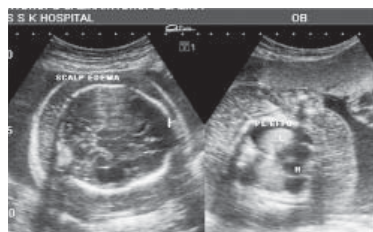


Figure 1: Ultrasound image of hydrops fetalis

Hydrops fetalis is a symptom rather than a diagnosis and the end stage of a wide variety of disorders. With timely diagnosis and improved treatment of Rhesus isoimmunization, nonimmune factors have become more frequent causes of hydrops fetalis. The incidence of nonimmune hydrops fetalis (NIHF) is estimated at 1 in 3000 pregnancies<sup>1</sup>. In NIHF 63% of cases belong to the five main categories: cardiovascular, chromosomal, thoracic, twin-twin transfusion syndrome (TTTS), and anemia<sup>2</sup>. Infectious and metabolic diseases can also cause NIHF<sup>2-3</sup>. NIHF has a favorable outcome in 27.5% of cases<sup>2</sup>, with idiopathic NIHF carrying the worst prognosis. Finding the etiology of NIHF is important for treatment and counselling regarding recurrence risk.

## Approach to A Case with NIHF

*All antenatal women with fetal hydrops should be referred promptly to a tertiary care centre for evaluation [IIA]<sup>4</sup>.*

## Maternal History

Maternal age, parity, period of gestation, consanguineous marriage, singleton or multiple pregnancy, history of fever with rash in mother, history of diabetes or hypertension in mother, history of previous child affected with same condition are some of the significant features to be elicited in history.

Table 1: Important points in history

Viral exposure/illness, medication
Ethnic background, consanguinity
3-generation pedigree
Fetal loss, death in infancy, developmental delay, congenital malformation, genetic syndrome
Underlying chronic illness- Preeclampsia (mirror syndrome), Sjogren lupus, Uncontrolled diabetes, Grave's disease

## Clinical Examination

A general examination of all such patient should be done at first visit. Important points to note while examining such patients are pallor, blood pressure, pedal edema, blood glucose levels. While doing per abdomen examination besides the routine parameters, one should look for the amount of liquor and twin pregnancy.

## Investigations

For the diagnosis of hydrops both invasive and non-invasive investigations are required. Many advanced investigations have come up which have made it possible for diagnosis of rare causes of non-immune hydrops. Also, the early diagnosis helps the family and the doctors to plan an action of management.

*Imaging studies should include comprehensive obstetrical ultrasound (including arterial and venous fetal Doppler) and fetal echocardiography. (II-2A)*

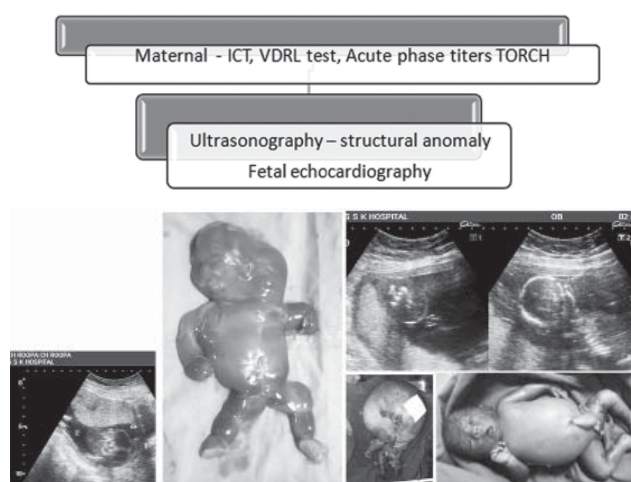


Figure 2: the ultrasound and postnatal picture of cystic hygroma and hydrops (left), twin reverse arterial perfusion (TRAP) with hydrops (right)

## Ultrasonography

- **Basics:** Basic two- dimensional ultrasound is done for assessment of amount of liquor. Polyhydramnios with amniotic fluid index more than 25cm is commonly seen, placentomegaly is said to be present if the thickness is more than 4cm in 2<sup>nd</sup> trimester and more than 6cm in 3<sup>rd</sup> trimester.
- **Targeted ultrasound:** For detection of structural anomalies commonly associated with nonimmune

hydrops. A detailed targeted anomaly scan of fetal system is very crucial.

- **Doppler:** The peak systolic velocity of fetal middle cerebral artery helps in diagnosis of fetal anemia.
- **Fetal Echocardiography:** Since congenital cardiac defects form a major etiological group among nonimmune hydrops; fetal echocardiography proves to be a useful investigation. It can detect almost all structural anomalies as well as arrhythmias.

Table 2: Structural anomalies on ultrasound leading to NIHF

<b>Head, Neck and Spine</b> <ul style="list-style-type: none"> <li>• Intracranial AV malformation - Vein of Galen aneurysm</li> <li>• Hydrocephalus - periventricular calcification</li> <li>• Cystic Hygroma</li> </ul>
<b>Cardiac anomalies</b> <ul style="list-style-type: none"> <li>• Multiple severe structural cardiac defects</li> <li>• Cardiac arrhythmia</li> <li>• Cardiomegaly</li> </ul>
<b>Thoracic anomalies</b> <ul style="list-style-type: none"> <li>• Congenital pulmonary and airway malformation</li> <li>• Congenital high airway obstruction</li> </ul>
<b>Gastrointestinal and urogenital tract anomalies</b> <ul style="list-style-type: none"> <li>• Meconium peritonitis</li> <li>• Calcification in abdomen</li> <li>• Lower urinary tract obstruction</li> <li>• Urogenital system malformation</li> <li>• Hepatomegaly</li> </ul>
<b>Skeletal anomalies</b> <ul style="list-style-type: none"> <li>• Short - long bones</li> <li>• Fractures, poor mineralization</li> <li>• Absent movement of limbs</li> <li>• Contractures</li> </ul>
<b>Twin Pregnancy</b> <ul style="list-style-type: none"> <li>• Twin twin transfusion Syndrome</li> <li>• Twin reverse arterial perfusion</li> </ul>
<b>Placental</b> <ul style="list-style-type: none"> <li>• Hemangioma- hyperdynamic circulation</li> </ul>

### Ultrasound Doppler - Middle cerebral artery (MCA) peak systolic velocity (PSV)

Doppler measurement of the MCA PSV should be performed in all hydropic fetuses after 16 weeks of gestation. MCA peak systolic velocity values are compared using centile chart, the values more than 1.5 MOM for the gestational age signifies severe anemia.

*In case of suspected fetal anemia, fetal blood sampling and intrauterine transfusion should be offered rapidly. (II2A)*

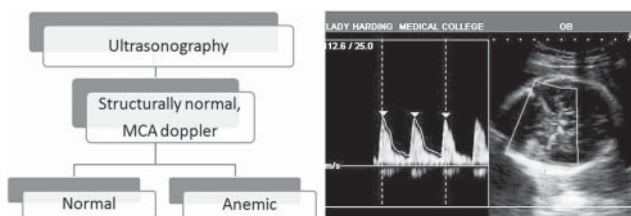


Figure 3: MCA PSV for the diagnosis of fetal anemia

### Causes of fetal anemia leading to hydrops

Table 3: Causes of fetal anemia in fetus and their diagnostic tests

α Thalassemia	Feto maternal hemorrhage
• Parental MCV - <80fL	• Kleihauer-Betke test
• Fetal blood smear - Bart Hb	• Flow cytometry
• Mutation detection by PCR	
Hemolysis	Parvovirus infection
• G6 PD deficiency by assay	• IgM antibodies in mother
• Pyruvate kinase deficiency	• Amniotic fluid PCR

*In unexplained fetal hydrops, investigation for maternal-fetal infections and tests for alpha-thalassemia in fetuses of women at risk because of their ethnicity should be performed (II-2A).*

### Invasive testing

*Fetal chromosome analysis and genetic microarray molecular testing should be offered where available in all cases of non immune fetal hydrops. (II 2A)*

### Amniotic fluid sample is to be sent for

- Karyotype/ chromosomal microarray (CMA)
- PCR - CMV, Parvo, toxoplasmosis
- Lysosomal enzyme testing
- DNA testing for specific anomalies as indicated

### Unexplained hydrops

All cases of unexplained fetal hydrops should be referred to a medical genetics service where available.

*Detailed postnatal evaluation by a medical geneticist should be performed on all cases of newborns with unexplained nonimmune hydrops. (II 2A).*

### Inborn errors of metabolism (IEM)

USG Features where IEM will be suspected

Severe fetal hydrops , increased NT
-Very thick skin, massive ascites
-Hepatosplenomegaly
-Contracture deformities, skeletal, renal abnormalities

### Diagnosis

- Measuring - specific lysosomal storage enzyme / metabolite, e.g Beta-glucuronidase deficiency in Mucopolysaccharidosis type VII, beta-galactosidase deficiency in GM1 gangliosidosis
- Histological examination of - liver, spleen, bone marrow, placenta
- Mutation identification is not easy. An index case is usually needed to confirm the diagnosis

## Treatment Modalities: -

*Some conditions such as fetal supraventricular tachycardia amenable to prenatal treatment represent a therapeutic emergency after 18 weeks. (II2A)*

Table 4: Disease condition and treatment modalities

Fetal anemia	Fetal blood sampling followed by in utero transfusion
Fetal arrhythmia	Medications such as digoxin, sotalol, propranolol, flecainide, amiodarone
Intrinsic thoracic malformations	Thoracentesis or thoracoamniotic shunt for pleural effusions in select cases
Twin twin transfusion syndrome	Fetoscopic laser ablation of communicating vessels
Syphilis	Penicillin

## Delivery in NIHF

Delivery should be done at tertiary care centre with facilities for appropriate speciality such as pediatric cardiologist in cardiac malformations, expert neonatologist in thoracic abnormality. There is no evidence that Caesarean section improves outcome. Cord blood can be sampled at delivery. Postmortem evaluation should be done in stillbirth/ neonatal death.

*Autopsy should be recommended in all cases of fetal or neonatal death or pregnancy termination. (II2A) Amniotic fluid and/or fetal cells should be stored for future genetic testing.*

## Tests after delivery

Neonatal / fetal demise	Neonatal survival
Clinical photograph	• Physical examination
Fetal tissue for further tests	• Echocardiography
• Fetal cells culture (skin, others)	• Cranial, abdominal USG
• Freeze fetal tissues and AF supernatant	• CBC, LFT, creatinine kinase, albumin, protein
• Bank fetal DNA	
Skeletal survey- Infantogram	• TORCH, viral culture
External examination and measurements	• Specialized testing - prenatal work-up
Internal examination - sending tissue for histopathology	
Examination of placenta/cord including histopathology	

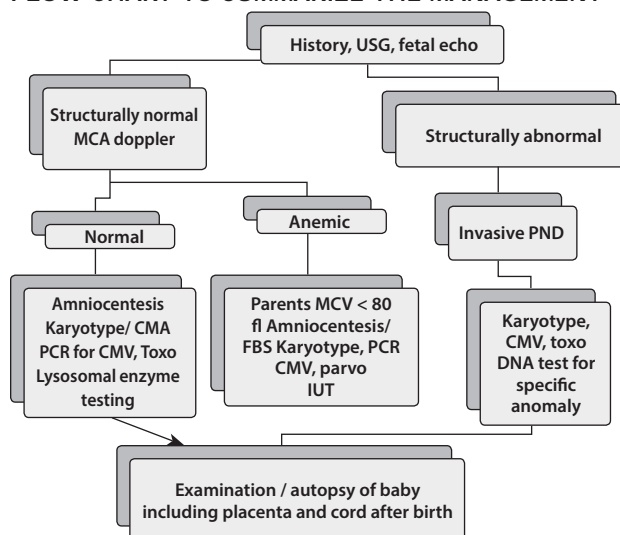
## Survival

Excluding chromosomal abnormalities, the survival rate of NIHF is about 10-30

- Structural heart defect - almost 100% mortality rate

Indicators of a poor prognosis
-Fetal chromosomal anomaly
-Gestational age < 24 weeks
-Fetal structural anomalies other than chylothorax
-Low 5-minute Apgar score
-Need for high levels of support during the first 24 hours

## FLOW CHART TO SUMMARIZE THE MANAGEMENT



## Our Experience

A prospective study was done in LHMC to evaluate the cause of NIHF cases referred to our centre and their outcome was analysed [5]. A total of 130 cases of fetal hydrops registered during eight-year study period were reviewed. Antenatal ultrasound, blood investigations and postnatal fetal examination were done, and outcome was noted. Out of 130 cases of NIHF, antenatal ultrasound showed the presence of structural malformations in 94/130 (72.3%), cardiac abnormality was the most common (34/130, 26.1%) and cystic hygroma was seen in 15/130 (11.5%). Chromosomal abnormality was observed in 15(11.5%) cases, and Doppler US showed anemia in 4/130 (3.1%) cases only. Live born were 25 (12.9%), and rest all were stillborn or abortion. Later mean gestational age of presentation ( $p = 0.0001$ ), presence of gastrointestinal malformation ( $p = 0.0001$ ) and absence of structural malformations ( $p = 0.0441$ ) were factors significantly associated with live birth; the presence of cystic hygroma ( $p = 0.0431$ ) or structural heart defect ( $p = 0.007$ ) was significantly associated with poor outcome. It was concluded that fetal anemia was not a common cause of NIHF in the study population. The early onset of hydrops and presence of structural malformation carry a graver prognosis; type of structural defect also has bearing on outcome.

## References

1. Bellini C, Hennekam RC. Non-immune hydrops fetalis: a short review of etiology and pathophysiology. Am J Med Genet A 2012;158A:597-605.
2. Randenberg AL. Nonimmune hydrops fetalis part I: etiology and pathophysiology. Neonatal Netw 2010; 29:281- 95
3. Bellini C, Donarini G, Paladini D et al Etiology of non-immune hydrops fetalis: An update. Am J Med Genet A. 2015; 167A(5):1082-8.
4. Désilets V, De Bie I, Audibert F. No. 363-Investigation and Management of Non-immune Fetal Hydrops. J Obstet Gynaecol Can. 2018 Aug;40(8):1091-1107
5. Kumar M, Jha V, Singh A. Nonimmune Hydrops Fetalis: Factors Which Predict outcome. J Obstet Gynaecol India. 2018 Jun;68(3):197-203.

# Managing Survivors of Sexual Violence

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The World Health Organisation (WHO) defines Sexual Violence as “any sexual act, attempt to obtain a sexual act, unwanted sexual comments/ advances and acts to traffic, or otherwise directed against a person’s sexuality, using coercion, threats of harm, or physical force, by any person regardless of relationship to the victim in any setting, including but not limited to home and work.” (WHO, 2003) Sexual assault is a form of sexual violence which is often used synonymously with rape. The Criminal Law Amendment Act (CLA) 2013 has expanded the definition of rape to include all forms of sexual violence-penetrative (oral, anal, vaginal) including by objects/weapons/fingers and non-penetrative (touching, fondling, stalking, etc.).

There are many health concerns of survivors/victims of sexual violence which need to be addressed. It is the duty of the state to ensure the right to health for these patients and provide appropriate physical and mental health services to them without discrimination. Health professionals have a dual responsibility towards these survivors. The first is to provide the required medical treatment and psychological support. Establishing a rapport with the survivor and showing empathy is of utmost importance. Secondly, they must assist survivors in their medico-legal proceedings by collecting evidence and documenting the findings according to the protocol.

## Standard Operating Procedure (SOP) for Management of Cases of Sexual Violence

Every hospital must have a Standard Operating Procedure (SOP) for management of cases of sexual violence. The SOPs must be printed and available to all staff of the hospital.

- Any registered medical practitioner can conduct the examination and it is not mandatory for a gynaecologist to examine such a case. In case of a girl or woman, every possible effort should be made to find a female doctor but absence of availability of lady doctor should not deny or delay the treatment and examination. In such a case, a male doctor should conduct the examination in the presence of a female attendant. In case of a minor/person with disability, his/her parent/guardian/any other person with whom the survivor is comfortable may be present.
- In the case of a transgender/intersex person, the survivor should be given a choice as to whether she/he wants to be examined by a female doctor, or a male doctor.

- Police personnel must not be allowed in the examination room during the consultation with the survivor. If the survivor requests, her relative may be present while the examination is done.
- Providing treatment and necessary medical investigations is the prime responsibility of the examining doctor. Admission, evidence collection or filing a police complaint is not mandatory for providing treatment.
- The history taking & examination should be carried out in complete privacy in the special room set up in the hospital for examination of sexual violence survivor. The room should have adequate space, sufficient lighting, a comfortable examination table, all the equipment required for a thorough examination, and the sexual assault forensic evidence (SAFE) kit for collecting and preserving physical evidence following a sexual violence.
- The collected samples for evidence may be preserved in the hospital till such time that police are able to complete their paper work for dispatch to forensic lab test including DNA.
- After the examination is complete the survivor should be permitted to wash up, using the toiletries and the clothing provided by the hospital if her own clothing is taken as evidence.
- Admission should not be insisted upon unless the survivor requires indoor stay for observation/treatment.
- Survivors of sexual violence should receive all services completely free of cost.
- A copy of all documentation (including that pertaining to medico-legal examination and treatment) must be provided to the survivor free of cost.

## Guidelines for Health Professionals

These guidelines laid down by the Ministry of Health & Family Welfare (MOHFW) describe in detail the stepwise approach to be used for a comprehensive response to the sexual violence survivor.

- Record the name of hospital where the survivor is being examined followed by the following details: name, address, age and sex of the survivor; date and time of receiving the patient in the hospital and commencement of examination; and name & relationship of the person who brought the survivor.
- Informed consent: A survivor may approach a health facility on her own only for treatment for effects of assault; with a police requisition after

police complaint; or with a court directive. In all circumstances, it is mandatory to seek an informed consent/refusal for examination; sample collection for clinical and forensic examination; treatment and police intimation. As per the law, the hospital/ examining doctor is required/duty bound to inform the police about the sexual offence and prepare a MLC. In case there is informed refusal for police intimation, then that should be documented. At the time of MLC intimation being sent to the police, a clear note stating “informed refusal for police intimation” should be made. The survivor or in case of child, the parent/guardian/or a person in whom the child reposes trust, has the right to refuse either a medico-legal examination or collection of evidence or both, but that refusal should not be used to deny treatment to survivor after sexual violence. Only in life threatening situations, the doctor may initiate treatment without consent as per section 92 of IPC. The consent form must be signed by the person herself if she is above 12 yrs. of age. Consent must be taken from the guardian/ parent if the survivor is under the age of 12 years. The consent form must be signed by the survivor, a witness and the examining doctor. Any major ‘disinterested’, person may be considered a witness

- **Relevant medical/surgical history:** It is important to record whether the survivor was menstruating at the time of assault/examination as some amount of evidence is lost because of menstruation and a second examination may be required on a later date in order to record the injuries clearly. Vaccination history is important with regard to tetanus and hepatitis B, so as to ascertain if prophylaxis is required.
- **Sexual violence history:** The doctor should record the complete history of the incident, in survivor’s own words as it has evidentiary value in the court of law. One must note who is narrating the incident-survivor or an informant. If history is narrated by a person other than the survivor herself, his/her name should be noted. Use of any Physical violence during assault must be recorded with detailed description of the type of violence and its location on the body (eg. Beating on the legs, biting cheeks, pulling hair, kicking the abdomen etc.). Information regarding attempted or completed penetration by penis/ finger/ object in vagina/ anus/ mouth should be properly recorded. Information regarding use of condom during the assault is relevant because in such cases, vaginal swabs and smears would be negative for sperm/ semen. In case of children, illustrative books, body charts or a doll can be used if available, to elicit the history of the assault. Post assault Information should be collected on activities like changed clothes, cleaned clothes, bathed/ urinated/ defecated/ showered/ washed genitals (in all cases) and rinsing mouth, drinking, eating (in oral sexual violence)/ had sexual intercourse after the incident of sexual violence. This would have a bearing on the trace evidence collected from these sites.

### *Examination and collection of samples for central/ State Forensic Science Laboratory (FSL)*

**Timing of evidence collection:** If a woman reports within 96 hours (4 days) of the assault, all evidence including swabs must be collected, based on the nature of assault that has occurred. Evidence on the outside of the body and on materials such as clothing can be collected even after 96 hours.

The nature of swabs taken is determined to a large extent by the history and nature of assault and time lapse between incident and examination. For example, if the survivor is certain that there is no anal intercourse; anal swabs need not be taken.

- **General Physical Examination:** Any signs of intoxication by ingestion or injection of drug/alcohol must be noted. Pulse, BP, respiration, temperature and state of pupils is recorded.
- **Examination for injuries and sample collection:** The survivor should be requested to stand on a large sheet of paper, so as to collect any specimens of foreign material e.g. grass, mud, pubic or scalp hair etc. which may have been left on her person from the site of assault/ from the accused. This sheet of paper is folded carefully and preserved in a bag to be sent to the FSL for trace evidence detection. Clothes that the survivor was wearing at the time of the incident of sexual violence are of evidentiary value if there is any stains/tears/trace evidence on them. Hence they must be preserved. (Always ensure that the clothes and samples are air dried before storing them in their respective packets. Pack each piece of clothing in a separate bag, seal and label it duly.) The entire body surface should be inspected carefully for signs of bruises, physical torture injuries, nail abrasions, teeth bite marks, cuts, lacerations, fracture, tenderness, any other injury, boils, lesions, discharge specially on the scalp, face, neck, shoulders, breast, wrists, forearms, medial aspect of upper arms, thighs and buttocks. The type of injury (abrasion, laceration, incised, contusion etc.) should be described including site, size, shape, colour, swelling, signs of healing, dimensions and simple/grievous. Injuries are best represented when marked on body charts. Swabs are used to collect bloodstains on the body, foreign material on the body surfaces seminal stains on the skin surfaces and other stains. Detection of scalp hair and pubic hair of the accused on the survivor’s body (and vice-versa) has evidentiary value. Collect loose scalp and pubic hair by combing. Intact scalp and pubic hair is also collected from the survivor so that it can be matched with loose hair collected from the accused. All hair must be collected in the catchment paper which is then folded and sealed. Nail clippings and scrapings must be taken for both hands and packed separately for DNA detection
- **Local examination of genital parts/other orifices:** External genital area and perineum is observed carefully for evidence of injury, seminal stains and

stray pubic hair. Any matted pubic hair should be clipped and sent for examination to detect the presence of semen. Two swabs are taken from the vulva, vagina, anal opening for ano-genital evidence. Two vaginal smears are to be prepared on the glass slide provided, air-dried in the shade and sent for seminal fluid/ spermatozoa examination. (Drying of swabs is absolutely mandatory as there may be decomposition/degradation of evidence which can render it un-usable.) Examination of the vagina of an adult female is done with the help of a sterile speculum lubricated with warm saline/ sterile water. A note is made of bruises, redness, bleeding and tears, which may even extend onto the perineum. In case injuries are not visible but suspected; we can look for micro injuries using a magnifying glass/ colposcope whatever is available. If 1% toluidine blue is available, it can be used to detect micro-injuries. Per speculum examination is not a must in the case of children/young girls when there is no history of penetration and no visible injuries. The examination and treatment as needed may have to be performed under general anaesthesia in case of minors and when injuries inflicted are severe. Per-Vaginum examination, commonly referred to by lay people as ‘two-finger test’, must not be conducted for establishing rape/sexual violence and the size of the vaginal introitus has no bearing on a case of sexual violence. Per vaginum examination should be done only in adult women when medically indicated. The status of hymen is irrelevant. An intact hymen does not rule out sexual violence, and a torn hymen does not prove previous sexual intercourse. One must refrain from commenting on the habituality of the survivor to sex based on status of the hymen. Genital findings must also be marked on body charts and numbered accordingly.

Bleeding/swelling/tears/discharge/stains/warts around the anus and anal orifice must be documented. Per-rectal examination to detect tears/stains/fissures/hemorrhoids in the anal canal must be carried out and relevant swabs from these sites should be collected.

Oral cavity should also be examined for any evidence of bleeding, discharge, tear, odema, tenderness. If history of oral penetration is present, oral swabs should be collected for detection of semen and spermatozoa. They should be taken from the posterior parts of the buccal cavity, behind the last molars.

- Investigations: Urine Pregnancy test should be performed and blood is collected for evidence of baseline HIV status, VDRL and HbsAg and sent to the hospital laboratory. If needed, radiographs of wrist, elbow, shoulders, dental examination etc. can be advised for age estimation. Blood and urine should also be collected for detection of drugs/alcohol and sent to FSL as the influence of drugs/ alcohol has a bearing on the outcome of the entire investigation. Venous blood is collected with the sterile syringe and needle provided and transferred to 4 sterile vials for

the following purposes: Plain Vial 1 - Blood grouping and drug estimation, Sodium Fluoride - Alcohol estimation, EDTA - DNA Analysis.

- Forming a provisional clinical opinion: Drafting of provisional opinion should be done immediately after examination of the survivor on the basis of history and findings of detailed clinical examination of the survivor.(Table)

Genital Injuries	Physical injuries	Opinion	Rationale why forced penetrative sex cannot be ruled out	What can FSL detect
Present	Present	There are signs suggestive of recent use of force/ forceful penetration of vagina/anus. Sexual violence cannot be ruled out.	Evidence for semen and spermatozoa are yet to be tested by laboratory examinations in case of penile penetration.	Evidence of semen except when condom was used
Present	Absent	There are signs suggestive of recent forceful penetration of vagina/anus.	Evidence for semen and spermatozoa are yet to be tested in case of penile penetration. The lack of physical injuries could be because of the survivor being unconscious, under the effect of alcohol/ drugs, overpowered or threatened. It could be because, there was fingering or penetration by object with or without use of lubricant - which is an offence under Sec 375 IPC	Evidence of semen or lubricant except when condom was used
Absent	Present	There are signs of use of force, however vaginal or anal or oral penetration cannot be ruled out.	The lack of injuries could be because of the survivor being unconscious, under the effect of alcohol/ drugs, overpowered or threatened or use of lubricant.	Evidence of semen or lubricant
Absent	Absent	There are no signs of use of force; however final opinion is reserved pending availability of FSL reports. Sexual violence cannot be ruled out.	The lack of genital injuries could be because of use of lubricant. The lack of physical injuries could be because of the survivor being unconscious, under the effect of alcohol/ drugs, overpowered or threatened. It could also be because, there was fingering or penetration by object with use of lubricant-which is an offence under Sec. 375 IPC	Evidence of semen, lubricant and drug/ alcohol

- Psychosocial care: All survivors should be provided the first line support. The health professional must provide this support himself/herself or ensure that there is someone trained at the facility to provide this.

- Treatment guidelines:

**Prophylaxis against Sexually transmitted infections (STI):** For non-pregnant women, the preferred choice is Azithromycin 1gm stat or Doxycycline 100mg bd for 7days, with Metronidazole 400 mg for 7days with antacid. For pregnant women, Amoxycillin/Azithromycin with Metronidazole is preferred. Metronidazole should NOT be given in the 1st trimester of pregnancy.

**Hepatitis B Prophylaxis:** A sample of blood for HBsAg is drawn and 0.06 ml/kg HB immune globulin is administered immediately (anytime upto 72 hours after sexual act).

Post Exposure Prophylaxis (PEP) for HIV should be given if a survivor reports within 72 hours of the assault. Before PEP is prescribed, HIV risk should be assessed.

**Pregnancy Prophylaxis (Emergency contraception):** The preferred choice of treatment is 2 tablets of Levonorgestrel 750 microgram, within 72 hours. If vomiting occurs, repeat within 3 hours. OR 2 tablets COCs Mala D - 2 tablets stat repeated 12 hours within 72 hours. Although emergency contraception is most efficacious if given within the first 72 hours, it can be given for up to 5 days after the assault. Pregnancy assessment must be done on follow up and the survivor must be advised to get tested for pregnancy in case she misses her next period.

**Tetanus toxoid (TT):** In case lacerations are present, assess whether the survivor is already immunized with TT. If not, administer ½ cc TT intramuscularly.

- **Follow-up:** The importance of follow up should be emphasized to the survivor. It is ideal to call the survivor for re-examination 2 days after the assault to note the development of bruises and other injuries; thereafter at 3 and 6 weeks. All follow ups should be documented. Test for pregnancy, STIs and assess for psychological sequelae and re-iterate need for psychological support.
- **Signature and seal:** After the examination the medical practitioner should document the report, formulate opinion, sign the report and handover the report and sealed samples to police under due acknowledgement. On the last sheet, mention how many pages are attached. Each page of the report should be signed to avoid tampering. It is important that one copy of all documents be given to the survivor as it is her right to have this information. One copy to be given to the police and one copy must be kept for hospital records. All evidence needs to be packed and sealed properly in separate envelopes. The responsibility for this lies with the examining doctor. All blood samples must be refrigerated until handed over to next in chain of custody. The hospital has the responsibility of properly preserving samples till handed over to police.

### *Special considerations*

- If a woman reports with a pregnancy resulting from an assault, she is to be given the option of undergoing an

abortion, and protocols for MTP are to be followed. The products of conception (PoC) may be sent as evidence to the forensic lab (FSL) for establishing paternity / identifying the accused. The examining doctor/AMO/CMO is to contact the respective police station, ask them to collect the DNA Kit from the FSL and bring it to the hospital to coincide with the time of MTP. The DNA Kit is used to collect the blood sample of the survivor. The accompanying DNA Kit forms are to be filled by the examining doctor. A photograph of the survivor is required for this form, and should be arranged for prior to the MTP. The products of conception (PoC) are to be rinsed with normal saline (NOT completely soaked in saline) and collected in a wide-mouthed container with a lid. This sample is to be handed over immediately to the police along with the DNA Kit, or preserved at 4 degree Celsius. It is to be transported by the police in an ice-box, maintaining the temperature at around 4 degree Celsius (2 to 8 degree Celsius) at all times.

- In most health centres because of the constant turnover, the doctor appearing in the court room could be different from the one who carried out the medical management of the survivor. In such instances, it is critical that the doctor making the court appearance be thorough with the case file of the survivor, such as, documentation of history examination findings and clinical inference drawn by the examining doctor.

### **Suggested Reading**

- Guidelines for medico-legal care for victims of sexual violence. Geneva, World Health Organization (WHO), 2003.
- Manual for Medical Examination of sexual assault by CEHAT 2010.
- The comprehensive health sector response to sexual assault. CEHAT working paper no.1, 2010.
- Guidelines & Protocols. Medico-legal care for survivors/ victims of Sexual Violence. 2014, Ministry of Health & family Welfare, Government of India.
- Manual for medical officers dealing with medicolegal cases of victims of trafficking for commercial sexual exploitation and child sexual abuse. Department of Women and Child Development, Government of India.
- A national protocol for sexual assault medical forensic examinations: adult/ adolescents. Washington DC, Office on violence against women, United States Department of Justice, 2004 (NCJ 206554)

## Events Held

- A Gynae Update on “Diagnosing Breast Lump” on 10<sup>th</sup> March, 2019 by IMA Janakpuri under the aegis of AOGD.



- CME on ‘Update on Ovarian Tumors’ under aegis of AOGIN and AOGD on 23<sup>rd</sup> March, 2019 at MAMC, LNJP Hospital.



- “FOGSI FORCE RAJDHANI”-PG Academic Programme on 26<sup>th</sup> & 27<sup>th</sup> March, 2019 at Lady Hardinge Medical College & SSK Hospital.



- Web telecast live RAJDHANI FOGSI FORCE- PG Academic Programme on 27<sup>th</sup> March, 2019 at LHMC



- Monthly Clinical Meeting on 5<sup>th</sup> April, 2019 at LHMC



- GBM & Handing over on 5<sup>th</sup> April, 2019 at Lady Hardinge Medical College.



- New Sub-committee Chairperson's Felicitation



# Menstrual Hygiene

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Dr Sumita Agarwal

Menstruation is a normal physiological process which is crucial for the sexual and reproductive health of women. Worldwide 52% of the female population is of reproductive age amounting to 1.9 billion women and adolescent girls menstruating each month. Menstrual Hygiene Management (MHM) is an integral part of menstrual health. MHM is defined by WHO and UNICEF (2014) as “Women and girls are using clean menstrual hygiene management material to absorb or collect blood, that can be changed in privacy as and when necessary for the duration of the menstrual period, using soap and water for washing the body as required and having access to facilities to dispose of used menstrual management materials.”<sup>1</sup>

Unfortunately, millions of women especially in low and middle income group countries like India still face significant barriers to a comfortable and dignified experience with menstrual hygiene management. It is difficult for girls and women to practice optimal menstrual hygiene because of inadequate information about puberty and menstruation, unavailability of appropriate WASH (water, sanitation and hygiene) infrastructure, and lack of access to hygienic menstrual management products which are either not available or unaffordable. This not only affects their health, well-being, and education, but also reinforces gender inequities and exclusion.

A study found that 71% of girls in India report having no knowledge of menstruation before their first period.<sup>2</sup> At menarche, schoolgirls in Jaipur, Rajasthan report their dominant feelings to be shock (25%), fear (30%), anxiety (69%), guilt (22%), and frustration (22%).<sup>3</sup> Further, 70% of women in India cannot afford to buy sanitary pads.<sup>4</sup> And according to a survey in 2012, 40% of all government schools lacked a functioning common toilet, and another 40% lacked a separate toilet for girls.<sup>5</sup>

Unsatisfactory menstrual hygiene management can have serious repercussions on a female's health in the form of increased risk of reproductive tract infections (RTI) like bacterial vaginosis or vulvovaginal candidiasis, urinary tract infections, infertility and psychosocial stress leading to depression. Few studies have shown that bacterial vaginosis (BV) may be more common in women with unhygienic menstrual hygiene management (MHM) practices.<sup>6,7</sup> In a recent case-control study in Odisha, India, it was observed that women of menstruating age with urinary tract infections (UTI) and BV were more likely using reusable absorbent pads than using disposable pads.<sup>8</sup> Both UTI and BV were also associated with a lack of a private space for changing, cleaning and washing during menstruation. As a girl progresses

from puberty into womanhood, RTIs potentially triggered by poor MHM could affect her reproductive health. Studies have shown women with BV may be at higher risk of adverse pregnancy outcomes like preterm birth, acquisition of sexually transmitted infections and development of pelvic inflammatory disease (PID).<sup>9,10</sup>

Since 2014, May 28 is observed as ‘Menstrual Hygiene Day’ worldwide to raise awareness regarding the challenges women and girls face to deal with menstrual cycles and highlight solutions implementable at global, national, and local levels to address these issues.

There is increased momentum from international donors, the national government, small and medium sized enterprises, and NGOs to address problems related to menstrual health. The focus to date has largely been on MHM products and improving awareness about menstruation. The various organizations are Goonj, Jayashree industries, Menstrupedia, UNFPA India, UNICEF India, WASH United India and WATERAID India to name a few.

## The basic enablers of a successful menstrual health program are

- 1) Education and awareness
- 2) Appropriate MHM Products
- 3) Sanitation
- 4) Government Policy

The relative importance and the manner in which these four interrelated factors manifest within a girl's life may vary and limitations in access to these can have long lasting effects on young women.

### 1) Education & awareness:

The majority of girls in India lack awareness about menstruation before menarche since it is a taboo subject and rarely discussed. Several regional studies have indicated that menstruating girls are not aware of the biological reasons associated with menstruation, and in fact perceived menstruation to be a “disease”. Girls as well as boys should receive accurate, timely information on the biological and psycho-social aspects of puberty, menstruation, and MHM from all three channels—mass media, influencers, and targeted education.<sup>11-13</sup>

### 2) Menstrual Hygiene Management Products

- A. Homemade products: Approximately 88% of women in India use homemade products (e.g., old cloth or rags) to manage their menstruation.<sup>14</sup> The main reasons for using cloth-based product are: personal preference and familiarity, lack of access to or affordability for high quality commercial sanitary pads, and lack of sufficient

information about pads. Some girls also use locally made cotton cloth. In a study of 164 adolescent girls in rural Gujarat, 68% said their first choice was a new soft cloth (falalin), while 32% said sanitary pads, and none of them preferred old cloths.<sup>15</sup> However, robust research on usage across India as well as impact on health outcomes has not been conducted.

- B. **Commercially available disposable pads:** Disposable sanitary pads are most common absorbents used especially in urban areas.<sup>16</sup> There are various health risks associated with the use of synthetic pads and tampons. It includes vaginal yeast infections and toxic shock syndrome (especially with tampon use). Also chemicals like dioxins and superabsorbent polymers (SAP) in these absorbents can accumulate and predispose to endometriosis, infertility and genital cancer.<sup>17</sup>

C. **Reusable pads:**

The demand for reusable cloth pads is low due to the upfront cost, lack of awareness about the product and its use and limited aspirational value.<sup>16</sup> Few organizations like EcoFemme in Tamil Nadu are manufacturing such pads.

- a. **Cloth pads:** Modern cloth pads are easily washable and come with modern conveniences like wings and leak proof layers. An ideal substitute for menstrual cups on low-flow days. For women who cannot use menstrual cups due to cultural taboos with virginity, a whole period packet of different sizes and absorbency would be ideal. Cost around 250 per pad which can be used for whole year. Reusable cloth pads also propagate the idea of “rash free, trash free, cash free.”
- b. **Inter-labia pads:** Inter-labia pads are worn between the labia to catch the menstrual flow and are used along with a cloth pad or a menstrual cup. They come in a leaf-like shape, are a lot smaller and thinner than a panty liner. They are designed to avoid leaks during heavy flow or flow with clots in it and can be worn along with a cloth pad. Inter-labia pads above pantyliners provide comfort, discretion, and a different way of catching flow before it leaks for those who tend to “gush”. These can also be used by women with mild stress incontinence.
- c. **Period panties:** They can be used alone or in conjunction with menstrual cups and come with removable inserts that can be changed and are leak-proof. Period panties comes with adjustable absorbency and are ideal for women with light, moderate or heavy flow.
- D. **Insertable products:** Insertable products (i.e., tampons and menstrual cups) are considered a high barrier product in India given apprehension among women with

inserting products, as well as the community’s perception that usage of tampons affects a woman’s virginity.<sup>18</sup> Leading stakeholders in MHM space including the national government do not advocate insertable products as an option for women and girls.

The menstrual cup is a small, bell-shaped cup usually made of medical-grade silicone that is inserted and worn inside the vagina during the menstrual cycle to catch and collect menstrual blood. Depending upon her flow, the woman removes the cup every 6-12 hours, empties the blood into the toilet, washes the cup and reinserts it into her vagina. One cup is all a woman needs and it can last her up to 10 years. There are different brand available in market and it costs around 850-1250 per cup. So it is user friendly, environment friendly and pocket friendly.

Barriers in the form of high price, poor access, and inappropriate use of preferred and appropriate MHM products are the most significant challenges facing women and girls in India. There is debate in the field about the appropriate MHM solution for women and girls in India given the environmental concerns about the increasing share of disposable sanitary pads. Although the national guidelines support the use of clean cloth, the Government of India is encouraging states to manufacture and increase access to disposable pads. The growing demand and supply of disposable sanitary pads present environmental concerns. In response, there have been some early efforts to develop innovative MHM products such as bio-degradable pads made out of locally grown materials such as bamboo, banana stem fiber, and sugarcane waste,<sup>19</sup> as well as re-usable cloth pads; however, these products are expensive and are not available at a large scale. Despite innovative methods to address the issue of last mile distribution through self-help-groups that decentralize production and distribution of pads, the need remains significant.

3) **Sanitation:**

There are 636 million Indians who lack toilets, and more than 72% of rural people practice open defecation;<sup>20</sup> lack of adequate sanitation disproportionately affects women. Without toilets in their home or public spaces, many women are forced to use public spaces to openly defecate and manage their menstrual needs. In addition to the impact on their health and dignity, women in communities face an increased threat of sexual harassment, rape, and other forms of violence.<sup>21</sup> Over the past decade, the Indian government, leading WASH donors, and NGOs have made significant efforts to build sanitation

infrastructure. They also recognize the need to drive behaviour change to encourage people to use toilets. More recently, leaders in the WASH space have begun to promote menstrual hygiene as fundamental to basic hygiene and sanitation services.

Limited access to functioning toilets remains a barrier and disproportionately impacts menstruating girls and women.<sup>22</sup> Access of toilet in the school, home and in communities should be provided. Even when toilets are available, cultural practices and hygiene routines as well as community attitudes related to menstruation limit the use of existing toilets, particularly during menstruation. Community attitudes and perceptions about menstruation and the availability of disposal infrastructure influences how women and girls dispose their menstruate waste.

#### 4) Policy:

Policies to improve MHM are led by multiple Ministries in India, with each Ministry bringing their own unique approach to address this cross-cutting topic. Overview of policies is depicted in Table1.

Table 1: Overview of policies in India relatrd to MHM

Ministry	MHM-related Policy or Program
Ministry of Health and Family Welfare (MoHFW)	The <i>Rashtriya Kishor Swasthya Karyakram</i> (RKSK), India's national adolescent health strategy, launched in January 2014 to prioritize access to MHM information, support, and MHM products through Adolescent Friendly Health Clinics and counselors.
Ministry of Drinking Water and Sanitation (MoDWS)	The <i>Swachh Bharat Mission</i> (SBM), India's national cleanliness program launched in October 2014, is run in rural areas by MoDWS and in urban areas by MoUD. It prioritizes sanitation infrastructure (e.g., individual and community toilets, solid waste management) and awareness programs for behavioural change. Recently, the MoDWS also took the leadership in drafting the National MHM Guidelines.
Ministry of Urban Development (MoUD)	
Ministry of Human Resource Development (MoHRD)	The <i>Sarva Siksha Abhiyan</i> (SSA, 2000-01) and <i>Rashtriya Madhyamik Shiksha Abhiyan</i> (RMSA, 2009), which aim to provide elementary education for all and enhance access to secondary education, respectively prioritize sanitation infrastructure in schools as a way to improve school retention. Additionally, <i>Swachh Bharat: Swachh Vidyalaya</i> (SB:SV), 174 India's national guidelines for sanitation in schools, emphasizes MHM facilities in schools(e.g., incinerators).
Ministry of Women and Child Development (MoWCD)	The <i>SABLA</i> program (2011), which is an integrated service to improve health, nutrition, and empowerment for girls, suggests providing awareness about MHM to adolescent girls through Anganwadi Centers.

Despite the array of initiatives put up so far, a myriad of challenges still remain to be addressed. Ensuring healthy

MHM for women would require a clear understanding of the intricacies related to the problem, the needs, and the influencing factors that could potentially affect the perception and practices of Indian women.

## Conclusion

Good menstrual hygiene triggers health, confidence, and self-esteem of women and is linked to gender equality and basic human rights. Concerns are manifold and calls for concerted multisectoral inputs and interventions to break the social taboos, myths, and misconception; support innovative sustainable solutions to manufacture and distribute low-cost, yet high-quality MHM products; and address the burgeoning problem of disposing menstrual waste in an environmentally safe manner. Ensuring menstrual hygiene for girls and women should be at the top of developmental agenda which calls for urgent and intensive action from all relevant stakeholders to change the scenario of menstrual hygiene in India. There is also emerging need for development of indicators under Swachh Bharat Mission Guidelines to measure the extent of achievement in MHM in India. Furthermore, setting up realistic time bound targets to indicate successful implementation of existing policy and programs would be a welcome endeavour for providing basic hygiene and reproductive services to girls and women.

## References

1. SHARE Consortium, London School of Hygiene & Tropical Medicine, Policy Brief, Menstrual Hygiene Management.
2. Mahon, Thérèse, Maria Fernandes. Menstrual Hygiene in South Asia: A Neglected Issue for WASH (Water, Sanitation and Hygiene) Programmes. London: WaterAid, 2010.
3. Gupta, Jaimala, Hitesh Gupta. Adolescents and Menstruation. The Journal of Family Welfare 2001; 47(1):1-13.
4. Sinha, Kounteya. "70% Can't Afford Sanitary Napkins, Reveals Study." The Times of India, January 23, 2011.
5. Bala, Nisha. "The 3 Biggest Reasons That India's Girls Drop Out of School." American India Foundation(blog), Summer 2014.
6. Baisley K, Changalucha J, Weiss HA et al. Bacterial vaginosis in female facility workers in north-western Tanzania: prevalence and risk factors. Sex Transm Infect 2009; 85: 370-375.
7. Balamurugan SS, Bendigeri N. Community-based study of reproductive tract infections among women of the reproductive age group in the urban health training centre area in hubli, karnataka. Indian J Community Med 2012; 37: 34-38.
8. Das P, Baker KK, Dutta A, Swain T et al. Menstrual hygiene practices, WASH access and the risk of urogenital infection in women from Odisha, India. PLoS One. 2015; 10(6): e0130777.
9. Nelson DB, Bellamy S, Nachamkin I et al. First trimester bacterial vaginosis, individual microorganism levels, and risk of second trimester pregnancy loss among urban women. Fertility and sterility 2007; 88: 1396-1403
10. Ness RB, Kip KE, Hillier S et al. A cluster analysis of bacterial vaginosis—associated microflora and pelvic inflammatory

- disease. American journal of epidemiology. 2005; 162: 585-590
11. Datta, Shib Sekhar, Nilratan Majumder. Sex Education in the School and College Curricula: Need of the Hour. Journal of Clinical and Diagnostic Research 2012;6(7):1362.
  12. Shanbhag, D., R. Shilpa, N. D'Souza, P. Josephine, J. Singh, BR Goud. Perceptions Regarding Menstruation and Practices during Menstrual Cycles among High School Going Adolescent Girls in Resource Limited Settings around Bangalore City, Karnataka, India. International Journal of Collaborative Research on Internal Medicine & Public Health 2012; 4(7): 1353-362.
  13. Sudeshna, Ray, Dasgupta Aparajita. Determinants of Menstrual Hygiene among Adolescent Girls: A Multivariate Analysis. National Journal of Community Medicine April-June 2012; 3(2): 294-301.
  14. Spot On! Improving Menstrual Health and Hygiene in India. Report. Dasra, Kiawah Trust, and USAID, 2014. 114
  15. Garg, Suneela, Tanu Anand. Menstruation Related Myths in India: Strategies for Combating It. Journal of Family Medicine and Primary Care 2015;4(2):184-86.
  16. Sanitary Protection in India. Country Report. Euromonitor International, June 2015
  17. Wendee Nicole. A Question for Women's Health: Chemicals in Feminine Hygiene Products and Personal Lubricants. Environ Health Perspect 2014 Mar; 122(3): A70-A75.
  18. Handique, Maitreyee. Fear of Losing Their Virginity Keeps Indian Women from Using Tampons. Quartz India, February 28, 2015.
  19. Rebello, Evans. Giving Her a Choice. The Kachra Project, May 28, 2015.
  20. Sanitation in India: The Final Frontier. The Economist, July 19, 2014.
  21. Gosling, Louisa, Chloe Irvine, Lisa Schechtman, and Yael Vellman. Nowhere to Go: How a Lack of Safe Toilets Threatens to Increase Violence against Women in Slums. WaterAid.
  22. Bala, Nisha. The 3 Biggest Reasons That India's Girls Drop Out of School. American India Foundation(blog), Summer 2014.

## Congratulations to Newly Elected AOGD Sub - Committee Chairpersons

2019-2021			
Sub-Committee	Chairperson	Contact No.	Email
Endometriosis committee	Dr Manju Khemani	9810611598	dr.manjukhemani@gmail.com
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2018-2020			
Sub-Committee	Chairperson	Contact No.	Email
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Rural Health Committee	Dr Abha Sharma	9868399727	drabhasharma.obg@gmail.com
Multidisciplinary Patient Sub-committee	Dr A.G. Radhika	9818065527	raradhikaag@gmail.com

AOGD members are invited to become members of various Sub-committee

Please contact respective Chairperson.

Membership of Maximum two Sub-committee can be taken at a time

# Approach to Psychosexual Problems in Adolescents



Dr Anjila Aneja

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## Introduction

“Adolescence” literally means “to emerge” or “to attain identity” and is essentially the period of rapid physical and psychological development starting from the onset of puberty to complete growth and development. Due to rapid urbanization, our way of living had changed completely over the period of time. Co-education, interaction through mobile and internet has resulted in greater social interaction between girls and boys. Indulging in sexual activity, commonly seen in western world is not so uncommon in Indian society at present times but there is relative low level of sex education amongst the adolescent’s due to cultural and social taboo. Due to lack of awareness and support from family and society, adolescents are commonly facing psychosexual problems leading to adverse consequence to the individual, family as well as the society. There are several initiatives that have been taken up by WHO to address and resolve these issues.<sup>1</sup>

Adolescent phase has been divided in three stages:

- Early adolescent: between 10 to 14 years of age. This phase include puberty, sexual explorations and insecurities about physical changes.
- Middle adolescent: between 14 to 18 years of age. This phase include building of thought process but impartial imagination of their action and may lead to early indulging in sexual activity and substance abuse.
- Late adolescent: 18 years and up. This phase has more maturity, empathy and understanding for consequence of own actions. they have broader sense of connection with their partners.

## Psychosexual issue in adolescents

Adolescent is phase of physical, mental and emotional development. Unacceptance or feeling of strangeness for such changes make these individuals stressful and depressive. Stress is more for a special subset of adolescent called sexual minoritized.

## Sexual minoritized

The term “sexual minoritized” encompasses a variety of gender and sexual identities and expressions that differ from cultural norms (eg, lesbian, gay, bisexual, transgender), as well as identities and expressions that defy discrete labels.

## Sexual orientation

It is pattern of physical and emotional arousal of genders to whom an individual is physically attracted.<sup>2</sup>

- Homosexual refers to sexual attraction for same sex individual
- Heterosexual refers to sexual attraction for opposite sex individual.
- Bisexual refers to sexual attraction for both sex individual.
- Pansexual refers to sexual attraction for any gender identity or biologic sex
- Asexual refers to lack of sexual attraction for any sex

**Sexual identity:** an individual assessment for his or her own sex orientation; those who have orientation for same sex are referred to as gay or lesbian.

**Sexual behavior -** Sexual behavior refers to particular sexual activities and incorporates the gender(s) of sexual partners. Youth who self-identify as heterosexual may engage in sexual activity with same-sex partners; youth who self-identify as homosexual may remain sexually inexperienced. Sexual behavior does not necessarily mean sexual orientation; it may represent experimentation, exploration, or exploitation.

**Body image-**Body image dissatisfaction is also quite common, more in school and college going girls. They do not hesitate to use adverse means to control weight. Unhealthy weight control behaviour including no eating for >24 hours, diet pill, vomiting, laxative and misuse of anabolic steroids is more common as compare to heterosexual peers.

In several large surveys, 4 to 8 percent of high school youth report same-sex attractions or behaviors.<sup>3</sup>

**Health issue and outcome related to sexuality in adolescent:**

Majority of sexual minoritized have no health issue but some of them have psychological and health problems. These youth have more risk of victimization, depression, suicide, substance abuse, homelessness, STDs and unplanned pregnancy compare to heterosexual youth.

## Unawarness for Contraception

Most of pregnancy occur due to unawareness of contraception, difficulties with adherence and continuation. Suboptimal contraception is also quite

common due to infrequent sexual activity. Unprotected sexual intercourse leads to unplanned pregnancies, STDs and HIV.

## Unplanned teenage Pregnancy

Sexual behaviours may lead to unplanned pregnancy. Consequences of early and unplanned pregnancy are several-fold. Pregnant adolescents are more likely to delay seeking prenatal care and have higher rates of unfavourable birth outcomes, such as prematurity, infant mortality, and poor health and developmental outcomes. Teen pregnancies are at a greater risk of mortality and morbidity. Approximately 16 million girls aged 15-19 years and 2.5 million girls under 16 years of age give birth each year in developing regions.<sup>4</sup> Complications during pregnancy and childbirth are the leading cause of death for 15 to 19-year-old girls globally. Every year, some 3.9 million girls aged 15-19 years undergo unsafe abortions.<sup>5</sup>

## Menstrual issues

Most common problems faced by adolescent girls are menstrual disorders like dysmenorrhoea and premenstrual syndrome. Hygiene-related practices of women during menstruation are of considerable importance, as it may increase vulnerability to Reproductive Tract Infections (RTIs) / STDs (Sexually Transmitted Infections). Poor menstrual hygiene is one of the major reasons for the high prevalence of RTIs in the country and contributes significantly to female morbidity. Most of the adolescent girls in villages use rags and old clothes during menstruation, increasing susceptibility to RTI's.

## STDs and HIV

In 2016, 2.1 million adolescents were living with HIV and 260,000 became newly infected with the virus. The number of adolescents living with HIV has risen by 30% between 2005 and 2016. The number of adolescents dying due to AIDS related illnesses tripled between 2000 and 2015, the only age group to have experienced a rise.<sup>6</sup> Infections including human papillomavirus, Chlamydia, and gonorrhoea have the highest rates in teens and young adults. Multiple factors that contribute to this increased risk including: early sexual activity, multiple sexual partners, inconsistent condom use and other barriers method, increased biologic susceptibility to infection, and difficulties accessing health care.

## Victimization

Sexual and psychological harassment that are quite common in adolescent age are: teen dating, rape, drug and alcohol abuse, suicidal provocation and forced to do unprotected sex. As Indian society is more male driven despite rapid urbanization, sexual harassment by

friends and close relatives are quite common and it is most often underreported due to lack of family support and cultural barriers. Victimization leads to school dropout, depression and increased suicidal tendency. Family rejection is very high in minoritized youth creating a vicious cycle of adverse psychosexual health. Minoritized youth have more risk of sexual prejudice, verbal harassment, physical and sexual abuse. They are harassed by family, peers and society.

## How to approach

**Sex education:** Education regarding responsible and healthy sexuality can and should be delivered at homes, schools, medical, and community settings.<sup>7</sup>

Schools have been an important source of mass sexual health education efforts like health goals, prevention of sexually transmitted infection [STI]/ human immunodeficiency virus [HIV], preventing unwanted pregnancy, abstinence, promoting condom use etc. In a meta-analysis of observational studies, parental monitoring was associated with delayed sexual intercourse, greater condom use and increased contraceptive use<sup>8</sup>. Learning about sex from friends, cousins, and the media is believed to be linked to increase the likelihood of engaging in sex. Media and technology also plays an important role in health and sex education.

The parents and family have an important role to play in healthy adolescent development. However, they may have difficulty accepting in a sexually minoritized child or family member due to fear of the unknown, fear of social stigma, and fear for their child's or relative's safety. This may be due to their own biases and personal, cultural, religious background. Parents should communicate to the child, both explicitly and implicitly, about their concern and affects on how the child views himself or herself.

Schools that institute specific policies and programs promoting safety and diversity create an opportunity for all students to achieve both academic and social goals. A tolerant and supportive school climate reduces harassment and victimization. Protective school environments has lowered the risks of substance use and suicidal tendency among sexual minoritized youth

**Barriers to sex education** – There are many barriers to discuss sexual issues with adolescents. These barriers include:

- Social taboo
- Lack of confidentiality
- Need for parental consent for any procedure related to contraception, STIs, may be a turn off for counselling.
- Personally biased conception of sexual health care due to political, religious, and ethical beliefs and biases
- Stigma and discomfort associated with various aspects

of sexuality including specific sexual behaviors, gender and sexual variance

- Cost effectiveness- Professional counselling is costly

Health care providers can help parents work through their values and beliefs in a way that consistently support's the child. Educating parent-to-parent, parents to child and support groups are important parts of counselling. Health care providers can guide parents and family in creating an accepting, comfortable and safe environment for their child.

It may be helpful and important to explain that same sex attractions are a normal variant of sexuality; is not a mental disorder .Familial support and acceptance of the sexual minoritized youth helps adolescents explore their sexual identities in a safe environment. Interventions that attempt to change sexual orientation (ie, "reparative" or "conversion" therapy) are ineffective, and potentially harmful (by increasing internalized stigma, distress, and depression).

## Principles of Care

Health care providers play an important role for adolescent health and sexuality.<sup>9</sup> Use of the following principles to guide discussions of adolescent sexuality help in individual strengths and responsibility

- **Confidentiality** - Assuring confidentiality is the first step in establishing basic trust and respect between the medical provider and the adolescent patient. Most adolescents ( lesbian, gay, bisexual, transgender, queer, or questioning (LGBTQQ)) youth require privacy to talk about their sexuality. Always ask partners, friends, or parents to leave the examination room before beginning these discussions. The importance of confidentiality in caring for adolescent (LGBTQQ), particularly surrounding issues of sexuality, cannot be overemphasized.
- **Normalization** - It is an important aspect of counselling. Health care providers should have communication style that helps them to ask the questions and get the answers they need to provide patients with appropriate health care, education, support, and counselling. Because many adolescents feel awkward and hesitate to answer questions which are sexual in nature, therefore the role of the clinician is to normalize discussions of sex.
- **Respect** - Health care providers must respect the diversity and differences inherent in adolescents and young adults, including gender, race/ethnicity, sexual orientation, and physical appearance. Respect is demonstrated through skilled communication with the use of open, nonjudgmental questions and avoidance of terms that could be perceived as pejorative such as "casual sex" or "multiple sexual partners"

Brochures, posters, pamphlets, and other information should be accessible.

- **Avoid assumptions** - It is important for health care providers to avoid making assumptions about the

sexuality of their adolescent clients. In particular, health care providers should not assume:

- That all patients are heterosexual unless told otherwise. Gay, lesbian, and bisexual youth exist in all communities and in all pediatric practices.
  - That males who self-identify as heterosexual do not have same-gender sexual partners.
  - That a self-identified lesbian does not require birth control. They can experiment with opposite-gender sexual partners and are at risk of becoming pregnant and acquiring STIs.
  - That female adolescents do not engage in anal sex.
  - That patients have all the information they need about safer sexual practices.
- **Specific questioning** - It is important for health care providers to ask specific questions

Specific sexual behaviors, including digital, oral, vaginal, and anal intercourse, date of last sexual activity and last sexual activity without a condom or without hormonal birth control measure. Sexual debut and lifetime, as well as recent numbers of partners, can be helpful components of a sexual risk assessment. Detailed information is important in determining risks and strengths.

- Contraception, for themselves or their partner. Questions about current use, prior methods, fears or concerns about side effects, and interest in other methods can generate useful discussion about contraceptive choices
  - Victimization, forced sex, or "sex against will" is common in adolescents and may be associated with other health issues including suicidal thoughts, substance use etc. Questions about exchanging sex for drugs, money, or housing may also be appropriate to ask to homeless and disadvantaged youth.
- **Listen to responses** - All patients deserve the undivided attention of their health care providers, especially when discussing sensitive topics. Adolescent clinics should provide elaborated counselling sessions as per their need .Timing of such clinics should be adolescent friendly, to avoid unnecessary school, college dropouts.
- **Avoid medical jargon** - It is important for health care providers use terms that are professional yet familiar and comfortable for the adolescent and avoid using medical jargon. As an example, for young men having sex with men, questions about anal sex may be replaced with more familiar terms, such as "top" or "bottom," respectively.
- **Recognize the links to sexuality** - an important factor is to recognize the connections between their sexual risk taking and other aspects of their lives. As an example, unsafe sexual practices while under the influence of drugs or alcohol, Other "links" include domestic violence, exchanging sex for money or drugs

- Advise prevention -Advices for healthy life style, quitting substance abuse, contraceptive practices , benefit of exercise, safe sexual practices, information about vaccination can be provided to young even while visiting for a sore throat, abdominal pain, or yearly anticipatory guidance. Each interaction can be seen by the health care provider as an opportunity for discussing health promotion and disease prevention.
- Know community resources - Adolescents may require or desire educational resources or other youth services. A variety of internet sites are available to provide relevant information and education on adolescent sexuality issues. They include:

American Academy of Pediatrics, American Social Health Association, National Youth Advocacy Coalition, Sex Information and Education Council of the United States, Numeours Foundation

## Summary

Sexuality, sexual behaviour and sexual relationships are an important and necessary part of adolescent development. One of the tasks for healthy adolescent development is the acquisition of a mature and responsible sexuality that leads to meaningful intimate relationships. "Responsible sexual behavior" (eg, delaying sexual intercourse upto a appropriate age ,involving with caring and respectful partners, increasing the use of condoms and other contraceptives) are important adolescent health issues. Health care providers are a valued and trusted source of information and advice for adolescent health and sexuality. It is important for them to utilize the opportunities to discuss sexual behaviour with adolescents by creating an open environment that leads to honest answers, focuses on individual strengths and goals, builds skills, and promotes personal sexual responsibility.

## Reference

1. Adolescent Friendly Health Services: An Agenda for Change. The World Health Organization 2004. [www.who.int/child\\_adolescent\\_health/documents/fch\\_cah\\_02\\_14/en/index.html](http://www.who.int/child_adolescent_health/documents/fch_cah_02_14/en/index.html) (Accessed on September 28, 2009).
2. Committee On Adolescence. Office-based care for lesbian, gay, bisexual, transgender, and questioning youth. *Pediatrics* 2013; 132:198.
3. Kann L, Olsen EO, McManus T, et al. Sexual identity, sex of sexual contacts, and health-risk behaviors among students in grades 9-12--youth risk behavior surveillance, selected sites, United States, 2001-2009. *MMWR Surveill Summ* 2011; 60:1.
4. UNFPA. Girlhood, Not Motherhood: Preventing Adolescent Pregnancy. New York: UNFPA; 2015. Available from: <http://www.un.org/en/development/desa/population/publications/dataset/fertility/adolescent-rate.shtml>. [Last accessed on 2018 Oct 08].
5. Darroch J, Woog V, Bankole A, Ashford LS. Adding it Up: Costs and Benefits of Meeting the Contraceptive Needs of Adolescents. New York: Guttmacher Institute; 2016
6. AVERT. Young People, HIV and AIDS; 2018. Available from: <https://www.avert.org/professionals/hiv-social-issues/key-affected-populations/young-people>.
7. Breuner CC, Mattson G, COMMITTEE ON ADOLESCENCE, COMMITTEE ON PSYCHOSOCIAL ASPECTS OF CHILD AND FAMILY HEALTH. Sexuality Education for Children and Adolescents. *Pediatrics* 2016; 138.
8. Dittus PJ, Michael SL, Becasen JS, et al. Parental Monitoring and Its Associations With Adolescent Sexual Risk Behavior: A Meta-analysis. *Pediatrics* 2015; 136:e1587.
9. Marcell AV, Burstein GR, COMMITTEE ON ADOLESCENCE. Sexual and Reproductive Health Care Services in the Pediatric Setting. *Pediatrics* 2017; 140.

## AOGD Sub Committee Nomination (2019-2021)

Nominations are invited for the post of chairperson of the following sub-committee for the year 2019-2021

✓ Oncology Committee

### Eligibility Criteria

1. Person should be a member of AOGD and have at least 10 years standing in the profession with at least 5 years duration of holding senior position in the respective institutions.
2. Chairperson of a subcommittee has to be a member of any subcommittee earlier for at least 1 year.
3. No repeat nomination will be considered after one term of two years.
4. In case of two people applying for the same post, the decision of the executive will be final.
5. In case of any deviation, the decision would be taken by executive committee.
6. Two posts cannot be held by any member at one particular time.

The nominations on plain paper should reach: AOGD Secretariat: Room No-307, Dept. of Obst & Gynae All India Institute of Medical Science (AIIMS) AOGD Office by 20th April, 2019 along with the bio-data stating the eligibility

# Domestic Violence

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Dr Niharika Dhiman

Domestic violence (DV) is a complex and multidimensional phenomenon considered as ill treatment towards human life.<sup>1</sup> Domestic violence is being considered as a public health problem, its consequences are damaging to the health and well-being of a women, as well as to the development of the child, family, community and society in general (WHO 2013).<sup>2</sup>

According to UN (United Nations) declaration, violence against women includes *"any act of gender-based violence that results in, or is likely to result in, physical, sexual or psychological harm or suffering to women, including threats of such acts, coercion or arbitrary deprivations of liberty, whether occurring in public or private life"*.<sup>3</sup> At a worldwide level, DV has reached large epidemic proportions, and the prevalence of violence during pregnancy has been described by researchers as situated within a range of 1-20%.<sup>4</sup>

Violence against women can be perpetrated throughout her life but violence during pregnancy harms the mother and the baby both physically and psychologically. Domestic violence (DV) is a term used in many countries, including low-and middle-income countries (LMICs), to refer to partner violence, but the term can also encompass child or elder abuse, or any other forms of abuse by any member of a household. The term Domestic Violence has been recently replaced by the term Intimate partner violence (IPV), which has been defined as: Behaviour by an intimate partner that causes physical, sexual or psychological harm, including acts of physical aggression, sexual coercion, psychological abuse and controlling behaviours. This definition covers violence by both current and former spouses and other intimate partners. Other terms used to refer to this include domestic violence, wife or spouse abuse, wife/spouse battering. Violence by a male intimate partner (also called "domestic violence"). IPV can affect women's health in three areas, physical, psychological and sexual.<sup>5,6</sup>

## Approach to a case of IPV<sup>5,6</sup>

### I. Identification of intimate partner violence

Two approaches have been described to enquire about IPV

1. *Universal screening or routine enquiry*
2. *Clinical enquiry or case-finding*- it is a more selective approach on the basis of clinical and diagnostic considerations. It is the identification of women experiencing violence who present to health-care settings, through use of questions based

on the presenting conditions, the history and, where appropriate, examination of the patient.

None of the methods have been found superior to the other. Though Universal screening has been implemented in some of High income group countries but has not been accepted worldwide.

Minimum requirements for asking about partner violence

- i. A protocol/standard operating procedure
- ii. Training on how to ask, minimum response or beyond
- iii. Private setting
- iv. Confidentiality ensured
- v. System for referral in place

### Identifying Warning signs of IPV

Women presenting with a vague symptoms such as: Asthenia, myalgias, headaches and migraines, menstrual disorders, shivering and hot flashes, digestive disorders and hypertension.

### Psychological warning signs are as follows:

Difficulty in concentrating, insomnia, nightmares, memory deficiency, difficulty making decisions, sadness, distrust of others and decreased self-confidence.

### Frequently asked questions to screen/ case finding of IPV:

- i. Has the partner slapped her, or thrown something at her that could hurt her
- ii. Pushed or shoved her
- iii. Hit her with a fist or something else that could hurt;
- iv. Kicked, dragged or beaten her up
- v. Choked or burnt her on purpose;
- vi. Threatened her with, or actually used a gun/knife or other weapon against her.

### Sexual violence has been identified by the following:

- Being physically forced to have sexual intercourse against will
- Having sexual intercourse because she was afraid of what her partner might do
- Being forced to do something sexual she found degrading or humiliating.

### Clinical conditions associated with intimate partner violence

- Symptoms of depression, anxiety, sleep disorders

- Suicidality or self-harm
- Alcohol and other substance use
- Unexplained chronic gastrointestinal symptoms
- Unexplained reproductive symptoms, including pelvic pain, sexual dysfunction
- Adverse reproductive outcomes, including multiple unintended pregnancies and/or terminations, delayed pregnancy care, adverse birth outcomes
- Unexplained genitourinary symptoms, including frequent bladder or kidney infections
- Repeated vaginal bleeding and sexually transmitted infections
- Chronic pain (unexplained)
- Traumatic injury, particularly if repeated and with vague or implausible explanations
- Problems with the central nervous system - headaches, cognitive problems, hearing loss
- Repeated health consultations with no clear diagnosis
- Intrusive partner or husband in consultations

*[Adapted from: Black MC. Intimate partner violence and adverse health consequences: implications for clinicians. American Journal of Lifestyle Medicine, 2011, 5:428-439.]*

## II. Create a Safe Environment for Assessment and Disclosure

Offer First-Line Support when women disclose violence. Always Discuss the Limits of Confidentiality Prior to Assessment.

First-line support includes: ensuring consultation is conducted in private

- ensuring confidentiality, while informing women of the limits of confidentiality
- being non-judgemental, supportive and validating what the woman is saying
- providing practical care and support that responds to her concerns
- asking about her history of violence, listening carefully, but not pressuring her to talk (care should be taken with the use of interpreters for sensitive topics)
- helping her access information about resources, including legal and other services that she might think helpful
- assisting her to increase safety for herself and her children, where needed
- providing or mobilizing social support.

If health-care providers are unable to provide first-line support, they should ensure that someone else (within their health-care setting or another that is easily accessible) is immediately available to do so. Follow-up consultation should be offered at an interval of 3-5 days and at 3 months.

Vulnerable groups such as mentally challenged or pregnant women have special requirements.

Offer first-line support to women survivors of sexual assault by any perpetrator, which includes:

- Providing practical care and support, which responds to her concerns, but does not intrude on her autonomy
- Listening without pressuring her to respond or disclose information
- Offering comfort and help to alleviate or reduce her anxiety
- Offering information and helping her to connect to services and social supports.

Take a complete history, recording events to determine what interventions are appropriate, and conduct a complete physical examination (head-to-toe including genitalia).

The history should include:

- The time since assault and type of assault
- Risk of pregnancy
- Risk of HIV and other STIs
- Mental health status.

Provide written information on coping strategies for dealing with severe stress (with appropriate warnings about taking printed material home if an abusive partner is there).

Psychological debriefing should NOT be used.

- Consider HIV PEP
- Offer emergency contraception where required
- Offer STI treatment
- Psychological support up to 3 months post-trauma
- Interventions from 3 months post-trauma

## III. Interventions

Various interventions have been developed for reducing DV and improving health outcomes.

### i. Psychological/mental health interventions

- a) Cognitive behavioural therapy (CBT): CBT is based on the concept that thoughts, rather than external factors such as people or events, are what dictate one's feelings and behaviour. CBT typically has a cognitive component (helping the person develop the ability to identify and challenge unrealistic negative thoughts), as well as a behavioural component.
- b) Eye movement desensitization reprocessing (EMDR): This therapy includes standardized procedures that include focusing simultaneously on (a) spontaneous associations of traumatic images, thoughts, emotions and bodily sensations, and (b) bilateral stimulation, most commonly in the form of repetitive eye movements.

### ii. Advocacy/Empowerment interventions:

- a) Advocacy - support that includes: provision of legal, housing and financial advice; facilitation

of access to and use of community resources such as refuges or shelters; emergency housing; informal counselling; ongoing support; and provision of safety planning advice

- b) Empowerment- Helping women to feel more in control of their lives and able to take decisions about their future,

iii. Mother-child interventions

- iv. Other interventions (expressive writing and yogic breathing).

IV. Special Groups:

- i. Pregnant women who disclose intimate partner violence should be offered brief to medium-duration empowerment counselling (up to 12 sessions) and advocacy/support, including a safety component, offered by trained service providers where health systems can support this. The extent to which this may apply to settings outside of antenatal care, or its feasibility in low- or middle-income countries is uncertain. IPV has a dual relationship with pregnancy. Associated adverse pregnancy outcomes including LBW, PTB, and neonatal death. Trauma can cause premature rupture of membranes or abruption of the placenta and subsequently PTB and LBW. There is a five times more likely to experience LBW
- ii. Where children are exposed to intimate partner violence, a psychotherapeutic intervention, including sessions where they are with, and sessions where they are without their mother, should be offered, although the extent to which this would apply in low- and middle-income settings is unclear.
- iii. Women who have spent at least one night in a shelter, refuge or safe house should be offered a structured programme of advocacy, support and/or empowerment.

V. Different models for delivering care for survivors of violence against women are as follows:

- i. Health care centres and clinics
- ii. District and regional centres
- iii. One stop centres

Such systems should be integrated into the present health care services for better identification and early detection of IPV cases. WHO does not recommend mandatory reporting of IPV but in case a women wants to seek police /legal support, reporting should be done.<sup>6</sup>

To conclude, WHO recommends that the following measures should be adopted by the healthcare system in order to address and prevent IPV<sup>2,6</sup>

1. Promote gender equality and women's human rights.
2. Establish, implement and monitor multisectoral action plans to address violence against women.
3. Enlist social, political, religious, and other leaders in speaking out against violence against women.
4. Data collection to monitor violence against women, and the attitudes and beliefs that perpetuate it.
5. Prevention of child abuse.
6. Integration of IPV with existing programmes for HIV and Adolescent health.
7. Provide safe environment for women and children at school.
8. Strengthen the government and non- government support system for women living with IPV.
9. Sensitize legal and justice systems to the particular needs of women victims of violence.

## References

1. Eustace J, Baird K, Saito AS, Creedy DK. Midwives' experiences of routine enquiry for intimate partner violence in pregnancy. *Women Birth*. 2016 Dec; 29(6):503-510.
2. WHO (2013) Global and Regional Estimates of Violence Against Women: Prevalence and Health Effects of Intimate Partner Violence and Non-Partner Sexual Violence. World Health Organization, Geneva.
3. United Nations. Declaration on the elimination of violence against women. New York, United Nations, 1993.
4. Audi et al. Violence against pregnant women: prevalence and associated factors. *Revista De Saúde Pública*, 2008; 42(5), 877-885.
5. Pallitto CC, García-Moreno C, Jansen HA, Heise L, Ellsberg M, et al. Intimate partner violence, abortion, and unintended pregnancy: results from the WHO multi-country study on Women's health and domestic violence. *Int JGynaecol Obstet*. 2013;120:3-9.
6. WHO multi-country study on women's health and domestic violence against women : summary report of initial results on prevalence, health outcomes and women's responses. World Health Organization (2005)

# Think before you.....Think !

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Dr Mohit D Gupta

“You are master of your unexpressed thought and slave of those expressed”

Human mind is the finest super computers. Each one of us is blessed with one. It can generate approximately more than 30,000- 50,000 thoughts per day. It has been studied that out these thoughts, only 1-2% are purposeful, positive and meaningful thoughts. Isn't that surprising? What about the rest? The other thoughts are negative, waste or purposeless thoughts.

Imagine owning the finest instrument in the world but not knowing to operate it, maintain it in the best possible way. This instrument is our mind. This is the most powerful tool that we have. It can create and automatically nurture what we put into it. It works in harmony with its other friend, the intellect. Let us understand power of mind.

- Mind is powerful machine: It is a powerful processor: it can generate any number of thoughts depending on my input.
- It works continuously: Right from our birth, mind functions continuously creating thought after thought. Even during sleep, our subconscious mind has the power to manifest thoughts.
- It creates our destiny inside before creating it outside: Whatever happens in our life happens twice: First it happens in our mind (thoughts) and then it manifests outside. It's our choice.
- It can transform and heal: If anything has to be changed in the conscious life, in the conscious world, it has to be first done inside.

Such is the power of this mind. Just as choosing a career is not taken lightly, I need to ask myself that how much do I value my thoughts when each thought is contributing to create my destiny.

Let us inspect how our life and feelings function today. If we face frequent anger, irritation, loss of purpose, depression, restlessness, hatred and jealousy, we have to just inspect our mind what kind of thoughts are we creating. It is surprising that our conscious life is in majority driven by those thoughts that have no aim or purpose.

These feelings can be transformed once we consciously start choosing our thoughts. This is optimal energy management.

The earlier paradigm was “think before you speak” and the newer paradigm is “Think before you think”.

Let us understand simple ways to optimal use of mind power.

1. Take responsibility: It is my energy and I have the choice to use it the way I want. Taking responsibility is an important and first step towards change. This helps me channelize the energy of my mind and determine what is happening in my life.
2. Monitor: We have a pool of inner resources. This includes our thoughts and time. If we waste them, there is a cost to myself. This happens when our thinking speeds up and we get stuck in perpetual thinking. Analysis and reanalysis will only harm and drain my energy. Let us monitor what goes on in my mind.
3. Pause frequently: Start with stopping frequently and taking a step back from the frequent, crazy speed of life. Touch base with your inner core to remember who you really are. Taking out time means to carry out a check on QUALITY and QUANTITY of thoughts.
4. Transform the input: What goes in will get processed and come out. If we exercise the muscles of our intellect by studying spiritual and positive knowledge early in the morning, we naturally start creating positive, powerful and purposeful thoughts. These thoughts stay with us.
5. Create Balance: Do everything in balance. Balance in thinking is reflected in form of comfort, joy, happiness and easiness in life. Balance does not mean to work less, but balance means to maintain easiness in thoughts and giving more and regular time to self so that energy of mind is maintained and life flows.
6. Giving away waste: Creating an internal barometer to check what depletes our energy. Maybe gossiping, speaking unnecessarily and thinking about other people, these behaviors are common negative ingredients that we feed to our mind and that deplete us. It is my responsibility to look after my own conscious mind and create a sacred space within.
7. Do things consciously: Conscious actions means to do a karma with understanding. We need to create a simple routine with adequate opportunities to let go of the routine to let off steam. Have time to smile and talk.
8. Make energy efficient changes:
  - a. Cooking and eat in appositive state of mind. Spiritual energy impacts my mind and my food and water.
  - b. Let go off past: When my mind takes me into

past it drains my energy. The point of maximum energy is the present moment. We need to train ourselves to observe the mind, bring it into the now and then act. Make the conscious effort to practice this again and again.

- c. Create positive responses with clean heart. A negative mind stagnates the energy.
9. Think before you 'think': Every thought has an impact. This understanding tells me to check every thought that comes in my mind. Is this thought needed? Is it beneficial to myself and the world?

Thoughts send vibrations and leave an imprint on my subconscious. Am I recording the right thoughts? How can I have thoughts that will make me fly, remain beyond worry and feel free constantly? I am responsible and I have a choice.

10. Meditate: Just as we stop to refuel our car, similarly few minutes of silence and meditation is what we need every day to empower and refuel our mind. This is no more optional. If we have to maintain our calm and happiness, meditation is the way.

Wishing you a beautiful Mind

## Congratulations !!

Dr Anita Rajhoria and Dr Anu Handa for successfully answering the quiz and crossword of March issue

\* \* \* \* \*

### Answer: March Issue

#### Crossword

Down: 1. WHO, 3. Stricture, 4. Mendelson, 6. Vaginal, 8. PPCM, 9. VTE  
Across: 2. PMCS (Perimortem caesarean section), 5. ECMO, 7. Versapoint, 10. Stenting

#### Pictorial Quiz

Figure 1: Ans 1. Transvaginal NOTES (natural orifice transluminal endoscopic surgery)

Ans 2. Tubal sterilisation, oophorectomy, salpingectomy, hysterectomy etc.

Figure 2: Ans 1. Extracorporeal membrane oxygenation, or ECMO

Ans 2. It is an advanced life support technique used for patients with life-threatening heart and/or lung problems



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## Clinical Utility of Expanded Carrier Screening: Reproductive Behaviors of At-Risk Couples

Ghiossi CE, Goldberg JD, Haque IS, Lazarin GA, Wong KK

### Abstract

Expanded carrier screening (ECS) analyzes dozens or hundreds of recessive genes to determine reproductive risk. Data on the clinical utility of screening conditions beyond professional guidelines are scarce. Individuals underwent ECS for up to 110 genes. Five-hundred thirty-seven at-risk couples (ARC), those in which both partners carry the same recessive disease, were invited to participate in a retrospective IRB-approved survey of their reproductive decision making after receiving ECS results. Sixty-four eligible ARC completed the survey. Of 45 respondents screened preconceptionally, 62% (n = 28) planned IVF with PGD or prenatal diagnosis (PNDx) in future pregnancies. Twenty-nine percent (n = 13) were not planning to alter reproductive decisions. The remaining 9% (n = 4) of responses were unclear. Of 19 pregnant respondents, 42% (n = 8) elected PNDx, 11% (n = 2) planned amniocentesis but miscarried, and 47% (n = 9) considered the condition insufficiently severe to warrant invasive testing. Of the

8 pregnancies that underwent PNDx, 5 were unaffected and 3 were affected. Two of 3 affected pregnancies were terminated. Disease severity was found to have significant association (p = 0.000145) with changes in decision making, whereas guideline status of diseases, controlled for severity, was not (p = 0.284). Most ARC altered reproductive planning, demonstrating the clinical utility of ECS. Severity of conditions factored into decision making.

### Editor's Comment

Expanded carrier screening is a method of screening where hundreds of recessive genes are analyzed in one test which helps in detection of couples at risk of transmission of a genetic disease to their child. The fallacy is that all the abnormalities detected may not have clinical relevance ie. not all mutations will produce an abnormal child in that couple and on the other hand it may cause unnecessary mental agony. Therefore expanded carrier screening may not be ideal under all circumstances and its role is yet to be defined.

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## Menstrual Hygiene, Management, and Waste Disposal: Practices and Challenges Faced by Girls/Women of Developing Countries

Rajanbir Kaur, Kanwaljit Kaur, and Rajinder Kaur

### Introduction

Menstruation wastes are the wastes that are generated by a female in her reproductive years. These wastes are produced during menstruation commonly known as menses, periods, or monthly bleeding cycle. The menstrual cycle has three phases, that is, follicular phase (proliferative), ovulation phase, and luteal phase (secretory). Menstruation is regulated by hormones; in this process, endometrium, lining of uterus, gradually thickens and sheds off and causes bleeding that normally last for 3-5 days and occasionally up to 7 days. Menstruation sheds two-thirds of the endometrial lining. In addition to blood, menstrual fluid contains mucus and vaginal secretions. The menstrual flow varies from female to female and may be more or less at the beginning of menses or may change throughout the cycle. The color of the menstrual fluid varies between

red, bright red, and dark brown to black. Menstrual fluid may or may not have unpleasant odour especially when it comes in contact with air. Menstrual flow or duration also changes before menopause or during gynaecological cancers. Under conditions of hormonal imbalance, fibroids, polyps, and endometriosis menstrual flow increase and excessive loss of blood through menstruation can lead to anaemia.

Women have developed their own personal strategies to handle this period of time. Globally, these strategies vary greatly due to the personal preferences, availability of resources, economic status, cultural traditions and beliefs, education status, and knowledge about menstruation. Practices related to menstruation hygiene are of major concern as it has a health impact; if neglected, it leads to toxic shock syndrome, reproductive tract infections (RTI), and other vaginal diseases

Poor genital hygiene negatively affects adolescents' health. Most girls are unaware and unprepared for menarche as they are not informed or ill-informed about menstruation. The main objective of this review was to summarize the concern and possible methods of menstrual waste management in low-income countries. The review article was aimed at understanding the menstrual practices, product design, demands, and disposal strategies. It includes both a summary of the existing menstrual hygiene needs and management and also an analysis of the current knowledge in the fields of public health, water and sanitation, and solid waste management.

## 2. Cultural Beliefs and Restrictions during Menstruation

Menstrual hygiene practices were affected by cultural norms, parental influence, personal preferences, economic status, and socioeconomic pressures. Menstrual beliefs refer to misconceptions and attitudes towards menstruation within a given culture or religion. Menstrual beliefs, knowledge, and practices were all interrelated to the menstrual hygiene management. By reviewing literature and articles published in journals and reports available on the Internet we found many cultural and religious beliefs followed by people regarding menstruation. These norms were the barriers in the path of good menstrual hygiene practices. Many women experiencing restrictions on cooking, work activities, sexual intercourse, bathing, worshipping, and eating certain foods. These restrictions were due to the overall perception of the people regarding menstruation as they consider it dirty and polluting.

In some parts of the country there were restrictions on bathing and a taboo against burial of bloodied menstrual cloth. Cloths should first be washed and then buried or reused. Washing and drying thought to be done secretly or in a hidden corner so that it cannot be seen by others. It was also believed that menstrual fluids may be misused for black magic, so women should wash the wrapper/cloth wore during menses only at night when others were asleep. Menstrual flow was seen as dirty, polluting, and shameful, so women hide menstrual cloths for fear of being cursed. In similar findings, it was believed that menstrual waste was linked to witchcraft and danger, so it must be buried unless witches go after human blood and find the menstrual wrapper/cloth and destroy the women by causing infertility. From all these beliefs, it was clear that education plays a key role in menstruation hygiene management. By educating both men and women regarding menstruation, we can overcome these false beliefs and taboos. Due to cultural expectations and restrictions many girls were not adequately informed about the realities of menstruation. As a result, they feel subnormal, diseased, or traumatized. Unprepared girls were frightened, confused, and feel embarrassed by menarche likely to develop negative

attitudes towards menstruation. Even touching of menstruating women was considered toxic, they were prohibited from cooking and from taking certain foods like pickle. These prohibitions are more in the rural areas than in the urban areas. They were also not allowed to participate in religious activities or to contact religious articles. Menstruating girls are also not allowed to bath and wash hair, as it is believed to impede blood flow.

## 3. Types of Absorbents Used during Menstruation

The preference of sanitary protection material is based on personal choice, cultural acceptability, economic status, and availability in local market. Along with basic sanitation facilities, one should be also provided with soap and menstrual absorbents to manage menstruation hygiene. The choice of absorbents varies among rural and urban women and girls. In rural areas, the most preferred absorbents are reusable cloth pads and in urban areas women prefer to use commercial sanitary pads. Chlorine-bleached Kraft or sulphate pulp is used by manufacturers to produce fluff pulp as absorbent used to make disposable sanitary products. Nowadays, many deodorised and non-deodorised sanitary products are available in the market made of synthetic fibre rayon. These deodorised products contain chemicals like organochlorines which have antibacterial activity. Due to their chemical composition, these products when buried in the soil they kill the soils microflora and delay the process of decomposition. Different menstrual products used by women/girls are discussed below (Figure 1).

### 3.1. Reusable and Washable Cloth Pads

They may be sustainable sanitary option but must be hygienically washed and dried in the sunlight. The sun's heat is a natural sterilizer and drying the cloths/cloth pads under it sterilizes them for future use. These cloth pads are reusable so they are cost-effective, easily available, and ecofriendly. They also need to be stored in a clean dry place for reuse to avoid contamination.

### 3.2. Commercial Sanitary Pads

They are easily available at many stores, chemist shops, or online. They are expensive, compared to cloth pads, nonreusable, and not very environment-friendly. The cotton used in their making is not 100% natural and may contain pesticides.

### 3.3. Tampons

They are the type of absorbent that provides internal protection. They are kind of plug of soft material (cotton) which is inserted into the vagina to absorb the menstrual flow before it leaves the body. They are expensive, not easily degradable in nature and, hence, not very environmental friendly. Nowadays, sea sponge tampons are available in the market which are a natural alternative to synthetic tampons.

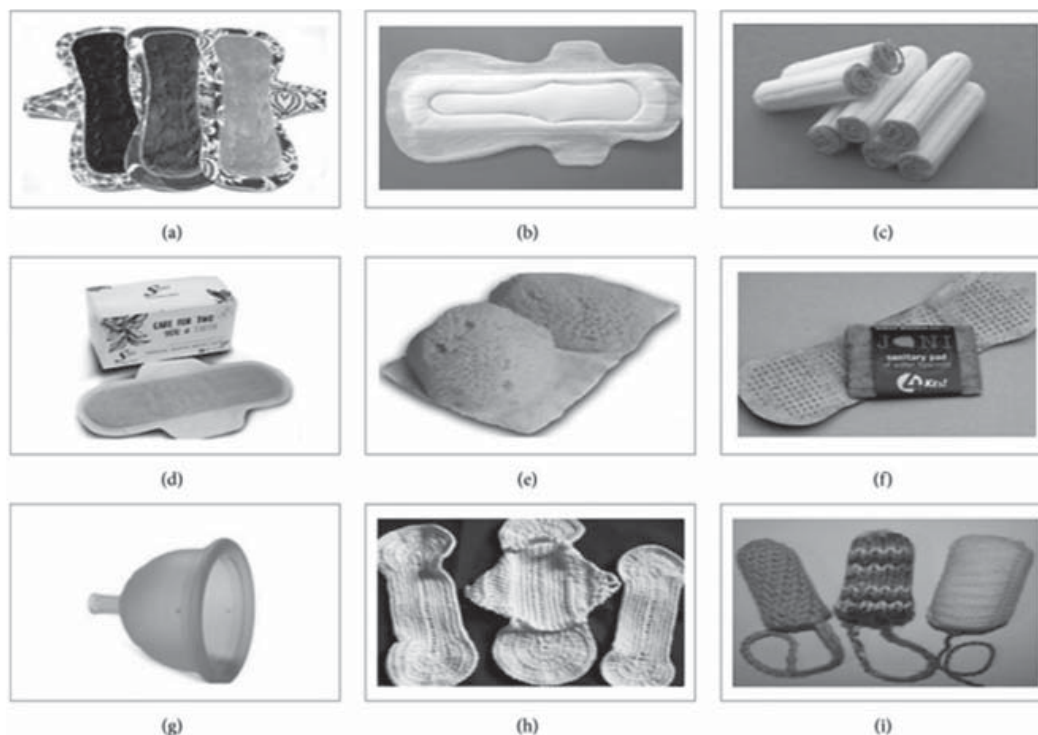


Figure 1

Types of sanitary products used by women during menstruation are (a) reusable cloth pads ([https://www.etsy.com/market/cloth\\_menstrual\\_pads](https://www.etsy.com/market/cloth_menstrual_pads)); (b) commercial sanitary pads (<http://topyaps.com/things-girl-must-know-about-sanitary-pads>); (c) tampons (<http://www.womensvoices.org/tag/tampons/>); (d) pads made from banana fibre (<https://saathipads.com/>); (e) sea sponges used as sanitary material (<https://www.pinterest.com/pin/194640015120225878/>); (f) pads made up of water hyacinth (<https://www.ecouterre.com/jani-a-biodegradable-sanitary-napkin-made-from-water-hyacinth/>); (g) menstrual cup (<http://rubycup.com/blog/how-to-clean-the-suction-holes-of-your-menstrual-cup/>); (h) pads made from wool (<https://www.pinterest.com/pin/198088083583361670/>); (i) reusable tampons (<http://naturalparentsnetwork.com/reusable-menstrual-products/>).

### 3.4. Reusable Tampons

These are washable tampons made up of natural materials like bamboo, wool, cotton, or hemp. They are also knitted or crocheted using the natural absorbent material like cotton or wool. They are inserted into the vagina to absorb menstrual flow same as the disposable tampons.

### 3.5. Menstrual Cups

They may be a new technology for poor women and girls and an alternative to sanitary pads and tampons. They are like cups made of medical grade silicone rubber which makes the cup easy to fold and get inserted into the vagina to collect menstrual blood. They can be worn up to 6-12 hours depending upon the amount of menstrual flow, so it needs to be removed and emptied less frequently. They are reusable and environment-friendly. It offers sustainable, practical, and cost-effective alternative where sanitation conditions are not good.

### 3.6. Bamboo Fibre Pads

Instead of wood pulp, bamboo pulp is used as an absorbing material in these sanitary pads. It has more absorbing capacity and is safer to use. They are affordable, easily decomposed, and environment-friendly pads which also possess antibacterial properties. This provides infection and irritation-

free menstruation. Also, bamboo charcoal pads are available in the market with advantage that blood stains are not clearly visible and are also reusable in nature.

### 3.7. Banana Fibre Pads

Nowadays, low-cost sanitary pads for rural women made from waste banana tree fibre were sold under trade name "Saathi" in India. They are environment-friendly and decompose within six months after use. Besides these products, women in the remote rural areas also use natural materials like cow dung, leaves, and mud (<https://sswm.info/category/background/background/background/health-and-hygiene-issues/menstrual-hygiene-management>).

### 3.8. Water Hyacinth Pads

Menstrual pads manufactured using water hyacinth is sold under trade name "Jani." They are cost-effective, easily biodegradable, and ecofriendly in nature.

## 4. Menstrual Waste Disposal Techniques Used by Women

Appropriate disposal of used menstrual material is still lacking in many countries of the world. Most of the countries have developed techniques to manage their fecal and urinary wastes but, because of lack

of menstrual management practices in the world, most of the women dispose of their sanitary pads or other menstrual articles into domestic solid wastes or garbage bins that ultimately become a part of solid wastes. Toilet facilities in India lack bins for the disposal of sanitary pads and hand washing facilities for menstruating women to handle menstrual hygiene. In urban areas, where modern disposable menstrual products are used they dispose of them by flushing in toilets and throwing in dustbins or through solid waste management, but, in rural areas, there are many options for disposing menstrual waste such as by burying, burning, and throwing in garbage or in pit latrines. In rural areas, mostly women use reusable and non-commercial sanitary materials like reusable pads or cloths. Thus, they generate lesser amount of menstrual waste as compared to women in urban areas who rely on commercial disposable pads. The menstrual material was disposed of according to the type of product used, cultural beliefs, and location of disposal. In slum areas, women dispose their menstrual waste into pit latrines as burning and burial were difficult due to limited privacy space]. The reason behind that is it was seen by men or used in witchcraft.

In schools, due to lack of sanitary facilities, girls throw their pads in toilets. In some cases, girls threw away their used menstrual clothes without washing them. Also many were reported being absent from school due to lack of disposal system, broken lock/doors of toilets, lack of water tap, bucket, and poor water supply. In some schools, incinerators or “feminine hygiene bins” are used for disposing menstrual waste material but due to shyness or fear of being seen by others they refrained from using it. The behavior of women regarding disposal is different when being at home and away from home. At home, they dispose the waste by wrapping and throwing in the dustbin along with other domestic waste. As mentioned above, the disposing habits change according to the place. In public places, prior to having knowledge about the consequences of flushing the pads, they flush them in the toilets or wrap and throw them in the dustbins. Where dustbins are not placed they leave the soiled pads wrapped or unwrapped in the toilet corners. This makes the toilets dirty, breeding place for flies and mosquitoes, and also unhygienic for other toilet users and cleaners. In many cities, the persons who manage the public toilets always complain of blockage of sewage system because of flushing of sanitary pads or rags in the toilet.

#### 5. Consequences of Menstrual Waste Disposal

As sanitation systems were designed with urine and feces in mind, they are unable to cope with the menstrual absorption materials. These absorption materials clog the sewer pipelines as they are unable to pass through and cause the system backflow. Materials like tampons, cotton wool, toilet paper, and other organic materials used for menstrual

management might be decomposed in pit latrines/landfills except the plastic inlay of the commercial sanitary pads. Sanitary napkins might decompose over a period of about one year except its plastic lining in on-site sanitation

In rural areas, pit latrines once full they were covered with soil and new pit was dug but due to space limitations this was not practiced in urban areas]. It was reported that some women and girls wrap their used menstrual cloths and packs in polythene bags before disposing in pit latrines which prevents them from decomposition. Nowadays, mostly women/girls prefer commercial sanitary pads and tampons which are made up of superabsorptive materials like polyacrylate. These pads and tampons when flushed in the toilets they get saturated with liquid and swell up, thus resulting in sewage backflow, a serious health hazard. The adhesive wings and the perforated plastic layers in the commercial sanitary napkins are not easily biodegradable. The sewage blockages were mostly due to accumulation of excessive quantity of solid waste or sand which results in hardening of the sludge in the pits. Blockage of sewage system is a global problem and major contributing factor is flushing of menstrual products in toilets. Deodorised sanitary products used by women/girls contain chemicals used in bleaching such as organochlorines which when buried in the soil disturb the soil microflora and decomposition takes time.

People living alongside river banks throw menstrual waste into water bodies which contaminate them. These materials soaked with blood were breeding places for germs and pathogenic microbes. Sanitary products soaked with blood of an infected women/girl may contain hepatitis and HIV viruses which retain their infectivity in soil and live up to six months in soil. The clogged drainage with napkins has to be unblocked and cleaned manually by conservancy workers with their bare hands without proper protection and tools. This exposes the workers to harmful chemicals and pathogens. Incineration is a better technique to dispose of menstrual waste but burning of pads releases harmful gasses that effects health and environment. Burning of inorganic material at low temperature releases dioxins which are toxic and carcinogenic in nature.

#### Strategies for the Management of Menstrual Waste

1. Disposal of menstrual waste is of major concern as it affects health and environment. There is a need for effective menstrual materials which needs less and cost-effective management.
2. Companies dealing with manufacturing of sanitary pads or other articles should disclose the information on the pads regarding the chemical composition of the pads so that appropriate technologies could be used for their disposal and treatment.

3. Environment-friendly chemicals should be used by manufacturers of sanitary products to stop soil and water pollution and to fasten the decomposition process.
4. Guidance regarding menstrual management to adolescent girls and women is a much needed step. Menstrual hygiene management should be an integral part of education curriculum.
5. Distribution of menstrual products should be free of cost in schools and educational institutes. Recently, instead of subsidizing the menstrual pads, Indian government has imposed 12% GST on them which is not very women friendly (source: <http://www.livemint.com/Industry/2Y4RRe0XaJmVduujmsDdXL/GST-rate-on-sanitary-napkins-fixed-at-12.html>).
6. The toilets must be designed and built to be girl/women friendly [45]. In Kerala, some schools have installed sanitary napkin vending machines in toilets which are semiautomatic and operate by inserting a coin in it. It contains 30-50 sanitary napkins to meet the emergency needs of the girls/women in schools (Figure 2).



Figure 2  
Sanitary napkin vending machine. Source: <https://timesofindia.indiatimes.com/city/pune/sanitary-napkin-vending-machines/articleshow/57824878.cms>.

7. There should be a separate collection system for the menstrual wastes without affecting the privacy and dignity of women. Specific sanitary dispensers to collect menstrual waste should be installed.
8. There should be sufficient space for washing, cleaning private parts and hands and for changing or dealing with stained clothes. To fulfil these requirements, there must be water availability, toilet paper, dustbin, and a sink to wash menstrual products.
9. Dustbins should be covered by lid and emptied from time to time to keep the toilets clean from flies, mosquitoes, and bad odour.
10. Covered containers and dustbins have advantage of hiding the waste being seen by others. They are installed in a place that offers privacy.
11. Gloves and proper safety tools should be provided to the cleaners so that they are not exposed to pathogenic organisms and harmful gasses.
12. Government should introduce new rules for the safe disposal and treatment of menstrual wastes as they have made for solid or biomedical wastes. Appropriate policy and legal framework is necessary for the management of menstrual wastes.

13. Government and non-government organizations should come forward for making the people aware of management of menstrual wastes. Government should give the funds to the Municipal Corporation or NGOs for the construction of women friendly toilets.
14. Health implications of menstrual wastes should be properly investigated. Financial support should be given to the institutions to carry out the research in the management of menstrual wastes.
15. Scientific research should be encouraged for the most suitable techniques of disposal of sanitary pads or other menstrual products.
16. Allocation of budget in schools to support menstrual hygiene management studies should be conducted.
17. Collaborative efforts (trash bins) should be made.
18. Incinerators are a better option for disposal but should be operated in a controlled environment so that harmful gasses emitted will not harm larger area.

### Better Ways/Ideas of Disposing Menstrual Wastes

#### Incinerators

If incinerators are used according to ecofriendly guidelines they create less pollution. They should be operated at certain specific temperature around 800°C so that they emit less harmful gasses. They should be installed in schools, institutions, and slum areas and at community level (Figure 3).



Figure 3  
Incinerator installed in the toilet for easy sanitary products disposal. Source: <http://www.vendingbiz.in/sanitary-napkin-incinerators-napkinci-maxi-1902444.html>.

#### Latrines with Chutes

These are special kind of toilets in which a shoulder level Chute was made in the usual deep pit. A chemical agent was added to the pit five times in a month to enhance the decomposition process of used napkins.

#### Reusable Cloth Pads

Using these reusable cloth pads is a better option as they have less chemical and plastic content. So they are easily decomposable as compared to other commercial products.

#### Biodegradable Products

Commercial sanitary product manufacturing companies must manufacture products having lesser chemical and plastic content. Pads made from bamboo fibre,

banana fibre, water hyacinth, and sea sponges should be encouraged.

#### Clay or Cemented Incinerators

Clay and cement incinerators used in Gujrat villages by “Vatsalya Foundation” are a welcomed step in menstrual hygiene management. A lady named “Swati” designed this incinerator and named it “Ashudhinashak” which burns many sanitary napkins at a time without creating any smoke. This ecofriendly and cheap innovation is appreciated by rural women who found difficulty in disposing them (Figure 4).

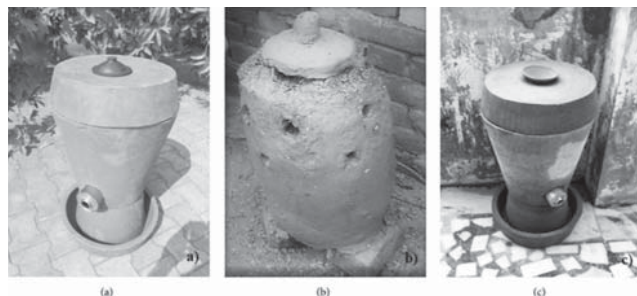


Figure 4

Incinerators used to dispose of menstrual waste in rural areas of India: (a) clay incinerator (<http://www.ecoideaz.com/innovative-green-ideas/ashudhinashak-clay-incinerators-for-sanitary-napkins>); (b) mud incinerator (<https://www.thebetterindia.com/87876/master-art-deal-with-menstrual-waste/>); (c) cement incinerator (<http://www.ecoideaz.com/innovative-green-ideas/ashudhinashak-clay-incinerators-for-sanitary-napkins>).

#### Better Disposal Techniques

Special covered bins should be installed to handle menstrual waste. Disposal bags should be provided by manufacturing companies with color indication for disposing these products. These bags should be freely distributed among schools and institutions. Menstrual waste should not be disposed of along with domestic waste. Pads should be properly wrapped in newspaper and then thrown in the dustbins. By this it should also be safe for rag pickers as it does not expose them to any disease-causing pathogens.

#### Conclusions

Menstrual hygiene should be promoted by implementing a course on menstruation and menstrual hygiene management. Teachers should be educated and trained to impart knowledge about menstruation and menstrual hygiene management among students. Social and electronic media also play an important role to make the girls and women aware about the latest menstrual products, different manufacturers, government policies, and so forth. Subsidies should be given on menstrual products so that every girl/women can afford them easily. Non-government organizations should come forward to educate rural people about menstruation, menstrual hygiene management, importance of toilets at homes, hand washing, diseases related to reproductive tract due to poor hygiene, and so forth. Emphases should be given on the use of reusable sanitary or cloth pads to overcome the problem of disposal. Girls and women should be aware of the consequences of disposing used menstrual products in open or flushing them in toilets. Dustbins with proper lids should be placed in the toilets. If possible, incinerators should be installed at homes, schools, and community levels. This study reveals that lack of privacy is a major concern both in household and in schools. Also, ignorance, misconceptions, unsafe practices, and illiteracy of the mother and child regarding menstruation are the root causes of many problems. So, there is a big need to encourage adolescents at school levels to practice safe and hygienic behaviors.

#### Editor's Comment

Menstrual hygiene and menstrual waste disposal is a real concern in India because most adolescents and women may not have the knowledge and access to safe menstrual practices. Providing low cost, easily biodegradable disposable pads or reusable cloth pads is the need of the hour. Access to free water and toilets is indispensable component of menstrual hygiene. Menstrual waste disposal with incinerators that emit less of harmful gasses or toilets with chutes which open into deep pits where pads can be degraded by adding chemicals may be the solution to the massive menstrual waste generated in our country.

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# Clinical Proceedings of AOGD Clinical Meeting held at Lady Hardinge Medical College, New Delhi on 5<sup>th</sup> April, 2019

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## Anhydramnios- Knocking the Wrong Door

Meenakshi Singh, Nihita Pandey,  
Ratna Biswas, Abha Singh

**Abstract:** Patients frequently present with early onset anhydramnios, 4 % of which remain unexplained. There remains a management dilemma regarding such patients in view of peri-viability. We present a case of early onset anhydramnios which turned out to be an intraligamentary pregnancy. Incidence of an intraligamentary pregnancy is 1 in 183000 pregnancies.

**Case Report:** A 30 yr old G2P1L1 with 24 weeks gestation was referred to a tertiary care centre in view of anhydramnios with low lying placenta. She did not have any complaints of leaking, pain in abdomen or fever. Her vitals and general examination were stable on admission. On abdominal examination, fundal height was 24 weeks, uterus relaxed with grossly reduced liquor, FHS present and regular. Her routine investigations were normal. USG Obs revealed an intrauterine fetus of 23 weeks, with nil liquor, no anomalies and type III placenta praevia. She was conservatively managed for 4 weeks, after which at 28 weeks gestation she developed fever and pain in abdomen. She had tachycardia of 120/min, per abdomen uterus 26-28 weeks size, tenderness was elicited and FHS 180/min. CRP was positive and TLC count 21,500/mm<sup>3</sup>. She was taken for caesarean in view of possible chorioamnionitis with placenta previa type III. Intra-op she was diagnosed as an Intra-ligamentary pregnancy in the Right broad ligament. Uterus was bulky and pushed postero-laterally towards the left side. A live female baby of 760 gms was delivered. Placenta was removed in bits and pieces. The sac was then sequentially clamped and excised. Right sided ovary which was adherent to the sac was salvaged. Post-op period was uneventful.

**Review of Literature:** Intra-Ligamentary pregnancy is a rare entity and accounts for 1 in 300 ectopic pregnancies. Multiple reports have been published but there is as yet no consensus on management. Of the 30 cases reviewed for our case, 21 cases presented in the first trimester, 3 presented in second trimester and 6 during the third trimester. Only one case could be diagnosed pre-op. The diagnosis was otherwise mostly during the intra-op period. Three live births have been reported so far. Removal of placenta needs to be individualised, since the attachment of placenta is important and its removal can lead to torrential bleeding. Only one case report mentioned leaving the placenta in-situ, whereas

all others could remove it without much complications. The ovary had been sacrificed in all the advanced broad ligament pregnancies, but we demonstrated that it can be salvaged and all attempt must be made in lieu of the same.

**Conclusion:** A TVS scan is important for diagnosis. A high index of suspicion should be maintained for patients who have a displaced cervix, early onset anhydramnios and failed induction of labour. Placental position needs to be determined before removal in such a case because the placenta may be located over major retro-peritoneal vessels.

## An Unusual Presentation of Post Caesarean Sepsis

Anuradha Singh, Manju Puri, Rama Anand

**Introduction:** The increasing rate of caesarean sections is a matter of concern for both developed and developing countries. Caesarean delivery is the single most important risk factor for puerperal infection in immediate postpartum period. Although most often the surgery may be uneventful but there is always a risk of infection with all surgical procedures. Here we report a rare case of Post caesarean sepsis where consequent to severe infection the uterus sloughed off and was expelled per vaginum.

**Case Report:** A 25 years old P2L2 with previous 2 LSCS presented on day 56 of LSCS with complaints of fever, foul smelling discharge per vaginum and pus discharge from drain site on abdomen for 1½ month.

Patient underwent LSCS for transverse lie with term PROM two months back. An abdominal drain was inserted which was removed on day 7 and patient was discharged. Stitch removal was done on day 18. Patient developed fever, swelling and pus discharge from drain site. She was readmitted and incision and drainage was done. Around 200cc of pus was aspirated and patient was discharged. She redeveloped the same symptoms and was referred to a higher centre.

On admission patient was febrile, pale, PR 120/minute, BP 100/60mm Hg. Per abdominally an ill-defined tender mass with variegated consistency was palpated in lower abdomen in the midline extending to right lumbar region corresponding to 18 weeks uterine size. On per speculum examination an irregular necrotic mass 3 cm in diameter with purulent foul smelling discharge was seen in upper vagina. On per vaginum

examination cervix was effaced forming a ring at vault; uterus upright, subinvolved, around 10 weeks size, adherent to abdominal wall, restricted mobility; an irregular mass protruding through os, upper limit could not be reached; left fornix clear, right fornix thickened; an ill defined mass 10x8 cm, felt to right of uterus in continuity with mass felt per abdomen. On pressing the abdomen pus drained through abdominal opening and vagina. Provisional diagnosis of chronic uterine inversion, placental polyp or fibroid polyp with PID and TO mass was made. Radiological investigations were suggestive of pyometra with a fistulous tract at drain site. Drain site pus culture revealed *Klebsiella*, sensitive to meropenem and colistin. Antibiotics were started with daily vaginal douching with diluted betadine. Patient improved but started complaining of dragging sensation in lower abdomen and something coming out of vagina. On examination a necrotic mass was seen protruding out of introitus. She was examined under anesthesia the mass could be pulled out completely from vagina followed by drainage of 100 ml of pus. On per vaginum examination after removal of mass, an opening felt at 11'O to 12'O clock position admitting 2 fingers, ? uterus felt in midline, normal size with restricted mobility. The histopathological examination of the mass revealed autolysed muscle. MRI on follow up (after 4 months of LSCS) revealed a very small atrophic uterus (5.4x1.5x2.3cm) with thinned out myometrium, ET 1.7mm, cervical stroma not visualised. Final diagnosis of Post caesarean uterine necrosis was made.

**Conclusion:** Caesarean section can be associated with serious short term and long term complications. Proper asepsis should be observed and any signs and symptoms of sepsis should be diagnosed early and managed aggressively. Promoting rationale and responsible use of antibiotics including the importance of single dose prophylactic antibiotic is essential

## A rare case of Recurrent Polyhydramnios

Manisha Kumar, Reena, Nishtha Jaiswal

**Background:** Polyhydramnios has incidence of 1-3/100 pregnancies[1]. It is defined as the pathological increase of amniotic fluid volume in pregnancy and is measured either as single deepest pocket > 8 cm, or the amniotic fluid index of more than 95% centile for the gestational age. The causes are fetal anomalies (8-45%), Gestational diabetes (5-26%), Fetal transplacental infections & anemia (1-15%), Multiple pregnancy (8-10%) [1].

**Case:** The patient was Mrs K, 23 yr old, non-consanguineous marriage, G2 P1L0, POG - 28 weeks, referred in view of polyhydramnios. She had no complains of breathlessness or pain abdomen. Her father was type II diabetes, she was on Tab Indomethacin (25mg) BD. In her investigations the CT

was negative, DIPSI was 149 mg/dl but her blood sugar profile was normal and Hb A1C was 4.6%, her VDRL test was non-reactive and TORCH serology was negative. The Indomethacin was continued and steroid cover was given. The Ultrasound(US) showed single live fetus of 28 weeks with AFI of 34 cm, no structural defect was identified, the bladder of fetus was consistently distended, amniocentesis for biochemical testing was done. Amniocentesis for biochemical analysis showed that the chloride levels were 121meq/l which was very high than normal of 108meq/l in amniotic fluid[2]. She was counselled that there was possibility of Bartter syndrome (BS) in the baby, the prognosis was guarded. The patient had respiratory discomfort at 32 weeks, amnioreduction was done and 3.5 liter was drained. The Indomethacin was stopped at 32 wks. She underwent amnioreduction three times until 36 weeks when she went into spontaneous labor. Controlled artificial rupture of membranes was done. She delivered fresh stillborn female baby of 2.06Kg. Cord blood was sent for next generation sequencing. The report showed homozygous mutation of KCNJ1 gene. She was counselled that the baby had BS type 2, which has antenatal presentation and is the severe form of BS. Chances of recurrence in next pregnancy was 25% as it is an autosomal recessive disorder (AR). Prenatal diagnosis can be done in the next pregnancy at 11-13 weeks by chorionic villous sampling and molecular testing for KCNJ1 gene mutation.

**Discussion:** Bartter's syndrome was originally described by Bartter and colleagues in 1962 [3]. The incidence is said to be 1: 50,000 - 1: 1,00,000 [4]. It is a renal tubular disease characterized by impaired salt reabsorption in the thick ascending limb of Henle's loop, resulting in excessive urinary losses of sodium, chloride, and potassium. There are 3 clinical variants namely neonatal, classic and Gitelman syndrome. BS results from mutations in numerous genes have mainly AR inheritance [4]. In the neonates it presents as prematurity, polyuria, and severe salt wasting and failure to thrive. The quality of life is poor, growth rate reduced, and hospitalization rate high. Numerous mutations have been identified. There was no known report of observation of constant full bladder in antenatal US which was found in our study.

**Conclusion:** This case brings into focus Bartter's Syndrome as a rare cause of unexplained severe polyhydramnios. The Red flags that should prompt suspicion of BS are unexplained, recurrent polyhydramnios, consistent full bladder on ultrasound.

## Fine Tuning Ultrasonography in Diagnosing Adenomyosis

Shreya Attri, Kiran Aggarwal, Sharda Patra

**Aims:** Evaluating the role of addition of doppler to greyscale ultrasound in differentiating clinically

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suspected cases of leiomyoma and adenomyosis of uterus and to correlate the assessment by ultrasound and Doppler findings with histopathological diagnosis.

**Method:** In this case series, 30 women with complaints of AUB and/ or dysmenorrhea were screened for a clinical diagnosis of adenomyosis or leiomyoma. This was followed by ultrasound and colour doppler. These patients were taken up for hysterectomy as planned and the uterus was sent for histopathological examination. Histopathology was taken as gold standard of diagnosis.

**Result:** Ill defined endometrial myometrial junction was the most common feature in women with adenomyosis

(90.9%). Clear demarcation of tumor margin (100%) was the most common morphological criteria in diagnosing leiomyoma. The presence of peripheral vascularity was seen in leiomyoma and central vascularity in adenomyosis. Addition of colour doppler to ultrasound was able to detect 27.7% additional cases of adenomyosis which were falsely labelled as leiomyoma by ultrasound alone.

**Conclusion:** Addition of colour doppler during routine ultrasound scan further enhances and fine tunes the diagnosis of adenomyosis and leiomyoma specially in young symptomatic women desirous of fertility.

## CROSSWORD

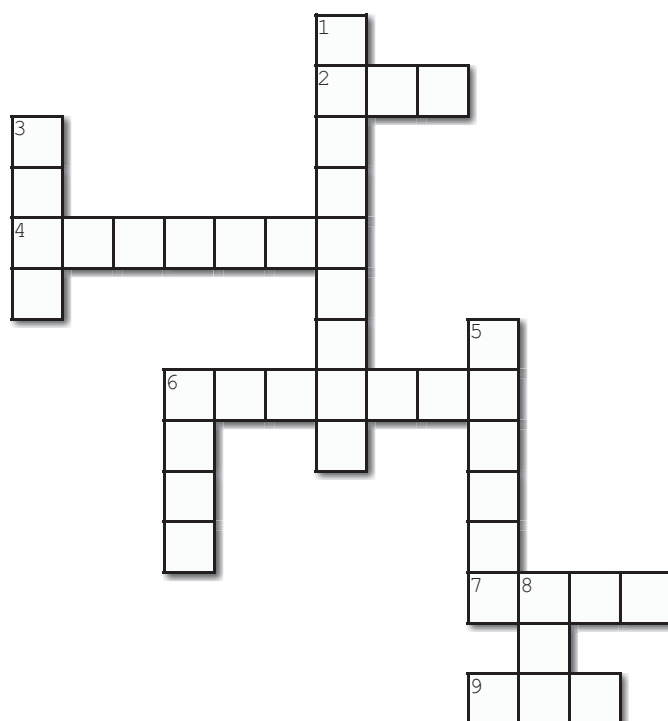
# The Maze of Knowledge

Swati Agrawal

Associate Professor, Department of Obs & Gynae, LHM & SSK Hospital, New Delhi



Dr Swati Agrawal



### Down

1. Drug associated with reduction in neural tube defects
3. Genetic profiling of embryos prior to implantation
5. Indian movie made on the issue of menstrual hygiene
6. Kit used for forensic evidence collection in survivors of sexual violence
8. Most common form of domestic violence

### Across

2. Centre dedicated to the care of survivors of sexual violence
4. USG done in second trimester to evaluate fetus for anomalies
6. Biodegradable low cost sanitary pad launched by the Government of India
7. Test to detect cell free DNA in maternal blood
9. Test done in early pregnancy to detect congenital abnormalities in the fetus

## PICTORIAL QUIZ

# A Picture is Worth a Thousand Words



Figure 1:

Q1. Identify the product & its use

.....  
 .....

Q2. List 2 advantages of this product

.....  
 .....



Figure 2:

Q1. Which procedure is being shown in the picture?

.....

Q2. Name any 2 complications associated with the procedure

.....  
 .....

WhatsApp your answers to 9953938995.

The names of first three correct entries will be mentioned in our next issue.

Refer page 43 for previous answer key.



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UK Faculty

**India Conveners and Contacts for details -**

Dr Nirmala Agarwal (n.menoky@gmail.com / 9811888732)  
Dr Arbinder Dang (arbidang@gmail.com 9871356917)

**For Accommodation, Hotel Bookings, Travel Enquiry Contact Miss Carolina Fernandez Cox & Kings +919711992043/  
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- Bank Transfer or Demand Draft must be made in favour of "RCOG NZ 2012 Plus" payable at New Delhi. (Cheques not accepted).
- There will be no refunds on cancellation.
- Registration request along with Demand Draft to be posted to the Secretariat mailing address as given below:-

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