Edition 3
November 2017

Healthy Babies to Healthy Mothers

ESTD 1989
ISPAT

# talkBACK

## Newsletter of the Indian Society for Prenatal Diagnosis and Therapy

### Meet our Team



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## editor's DESK

Dr. Usha Dave

Dear ISPATIAN Friends,

Wising you all a very happy and prosperous New Year and a great Diwali Festive Season!

The last few years have seen extraordinary advances in prenatal genetic diagnosis and practice led by many technological advances, be it next generation sequencing of cell free DNA in maternal plasma to non-invasively identify fetal chromosomal abnormalities or a microarray analysis of chorionic villous sampling and amniotic fluid samples, resulting in better cytogenetic resolution.

Dr. Seema Pandey

We at ISPAT, under the able guidance of our president, are trying to sensitize our fellow colleagues to be more conversant with all these latest modalities. Our approach is multipronged, at one level ISPAT is conducting a series of CMEs nationwide on the basic topics like prenatal diagnosis and the role of your USG machines. At another level multiple webinars are being organized to exploit the perks of advanced media and web services through which you are just a click away from the learning, totally hassle-free. This Newsletter is also a part of that teaching and training program, where we select one basic but contemporary topic to elaborate its intricacies in simple language. We try to keep you updated with upcoming events like interesting national and international conferences. A few pictures of past ISPAT events help you know about the happenings. Academically, we are trying to give expert views on a recent book launch or a breaking news in the field of fetal medicine & our quiz section is to stimulate your neurons.

The article of this month is about a very basic topic 'Pedigree Chart', how to make it and read it. This we thought would help you next time when you come across a patient with some complicated genetic trait or a colleague of yours has sent you a chart as her expert opinion as a

## president **MALHOTRA**

Dear ISPATIANS

Greetings & warm Regards

ISPAT is now being recognized as a progressive academic association of India. We are affiliated with World Association of Prenatal Medicine, Fetus as a patient, IAN DONALD SCHOOL of Ultrasound and International Academy of Prenatal Medicine.

Our members participated in the recent world congress at Belgrade and the ISPAT session was the most attended session in the congress.

Also a lot of ISPAT member gave lectures and did workshop in the recent world congress of IAN DONALD SCHOOL at Dubai.

On the national front our FGR-Genetic 6 CMES all over India were well revered. Our live webinar from Delhi in August got a record number of views.

We will continue into 2018 in a similar way with 6 city CME's on Genetics, Webinars and International participation.

We look forwards to many obstetricians joining ISPAT as life members and we also look forwards to new chapters.

LONG LIVE ISPAT

Dr. Narendra Malhotra
President

## Continued from **Pg1**

geneticist.

As the Editors of this e-Newsletter, our aim is also to get some feedback on our efforts so that we can improve in next issues. We would be more than happy if you come forward and share your experiences in the form of an interesting case, any article review or any innovative work or simply a genetic healthcare activity taken place in your area. In next few years, ISPAT aims to make its presence felt throughout the country so that we can learn from each other and apply it to our patients who can get benefitted by our combined efforts.

Wishing you all a happy reading.

**Let's salute their efforts-**

With the help of Rotary Club of Midtown, a Genetic Health Clinic is started with Free Genetic Counseling


services for rural community, at Kadamwadi, Kolhapur, Maharashtra. It is conducted by Dr. Usha Dave, Medical Geneticist & assisted by local pediatricians, Dr. P. Sanghavi & Dr. S. Kulkarni along with Rehab

## What's in **NEWS** these days

WGS Identifies New Genetic Signature for Autism:

In a breakthrough discovery, a complete new set of genetic signatures of the Autism has been found inside the cell. Autism a genetic disease, but most of the causes are unexplained till now. The results were published in September 2017 issue of the journal 'Cell'. This discovery will help physicians in treating these patients better if it could be replicated as a routine test.

*https://www.technologynetworks.com/genomics/news/wgs-identifies-new-genetic-signature-for-autism-293245*

## membership **UPDATE**

# NEW MEMBERS

| | |
|---|---|
| HARYANA | 33 |
| UP | 27 |
| CENTRAL | 2 |
| TOTAL | 820 |

**Become an ISPATian**

**https://goo.gl/ExJQvv**

## upcoming **EVENTS**


## What's in **NEWS** these days

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international **UPDATE**

## WORLD ASSOCIATION OF PERINATAL MEDICINE

### Belgrade Declaration on Healthcare Justice for Pregnant, Fetal, and Neonatal Patients

*Frank Chervenak1, Laurence McCullough1, Aleksandar Ljubic,2 Asim Kurjak,3 Milan Stanojevic4*
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*2Medigroup system, Belgrade, Serbia, DIU Libertas, Svetog Dominika 4, Dubrovnik, Croatia;*
*3Department of Obstetrics and Gynecology, Medical University of Zagreb, Zagreb, Croatia;*
*4Department of Obstetrics and Gynecology, Medical School University of Zagreb, Neonatal Unit, University Hospital "Sveti Duh", Zagreb, Croatia*

Global organizations have emphasized the need for healthcare for pregnant women and newborn children. The World Health Organization states: "Pregnancy – the nine months or so for which a woman carries a developing embryo and fetus in her womb – is for most women a time of great happiness and fulfillment. However, during pregnancy, both the woman and her developing child face various health risks. For this reason, it is important that all pregnancies should be monitored by skilled care providers." (http://www.who.int/topics/pregnancy/en/) The United Nations Convention on the Rights of the Child states that all children "…have the right to good quality health care – the best health care possible..." and that "Rich countries should help poorer countries to achieve this" (http://www.ohchr.org/EN/ProfessionalInterest/Pages/CRC.aspx). In response to these other international statements, campaigns for the improvement of maternal and newborn health have made progresses but much remains to be done.

The World Association of Perinatal Medicine is committed to protecting and promoting the health-related interests of pregnant, fetal, and neonatal patients worldwide. This is a vulnerable population of patients in many countries, especially low-income countries or after major natural disasters. Perinatologists in all countries should advocate for the resources needed to protect and promote the health-related interests of pregnant, fetal, and neonatal patients.

Ethical Framework

The ethical concept of healthcare justice creates an ethical obligation to provide clinical care that a patient needs in evidence-based clinical judgment. Healthcare justice creates the professional responsibilities of perinatologists in patient care and advocacy for health policy.

Professional Responsibilities in Patient Care of Pregnant, Fetal, and Neonatal Patients

- Develop clinical practices based on the best available evidence about maternal, fetal, or neonatal benefit of clinical management
- Prevent biases that arise from individual or group self-interest, to promote evidence-based practice
- Conduct scientifically, clinically, and ethically well-designed research to improve perinatal outcomes
- Recommend evidence-based clinical management
- Implement the healthcare-justice-based rights of pregnant, fetal, and neonatal patients to evidence-based clinical management
- Prevent clinical management that lacks an evidence base and should therefore be considered wasteful
- Implement informed consent processes that explain evidence-based clinical practice to pregnant patients and new parents, to gain their trust in evidence-based practice
- Implement culturally sensitive and competent decision making processes with pregnant patients and new parents
- Commit to an ethics of cooperation among professions and disciplines, with the goal of providing comprehensive, integrated clinical management for pregnant, fetal, and neonatal patients
- Educate patients and society that the prevention of diseases as well as promotion of health should begin in utero, because there is increasing evidence that prenatal development is a major determinant of adult health and disease

Professional Responsibilities in Advocacy for Health Policy for Pregnant, Fetal, and Neonatal Patients

- Advocate for public and private resources in one's own county that are required to deliver evidence-based clinical management in the appropriate institutional setting
- Advocate for healthcare policies that encourage

talkBACK

## international UPDATE

research and development to guarantee a constant, dynamic improvement in perinatal healthcare

• Advocate for recognition of the healthcare-justice-based rights of pregnant, fetal, and neonatal patients to evidence-based clinical management

• Advocate for health policy that supports perinatologists in preventing wasteful clinical practices

• Advocate for public and private resources from high-income countries that are required to deliver evidence-based clinical management in the appropriate institutional setting

• Advocate for inclusive process for decision making about national healthcare budgets so that the health interests of pregnant, fetal, and neonatal patients are addressed

• Advocate for health policy designed to prevent bias originating in individual or group self-interest, including appropriate use of the civil and criminal law to support evidence-based clinical practice

• Advocate for health policy that promotes an ethics of cooperation among professions and disciplines, with the goal of providing comprehensive, integrated clinical management to pregnant, fetal, and neonatal patients

• Advocate for resources to prevent clinically unacceptable differences in outcomes for pregnant, fetal, and neonatal patients that can be created by national and regional differences in perinatal biotechnology

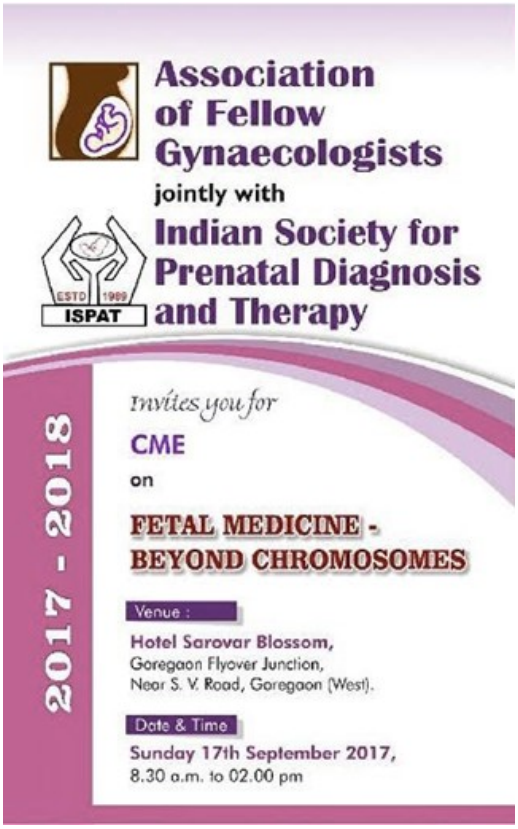
• Advocate for resources to prevent diseases and promote health of fetal patients, beginning in utero, in response to increasing evidence that prenatal development is a major determinant of adult health and disease


13th WCPM
2017
13th WORLD CONGRESS
OF PERINATAL MEDICINE
13th WCPM
World Congress of Perinatal Medicine


Narendra Malhotra
Milind Shah


ISPAT
SESSION
@
13th W.C.P.M Belgrade
27th Oct 2017

## some PAST EVENTS





## quiz of the MONTH

by Dr. Selvapriya

1. WITH FIRST CHILD HAVING NEURAL TUBE DEFECT WHAT IS THE BEST OPTION FOR NEXT PREGNANCE
   - A. Folic acid 4mg per day
   - B. Vitamin c
   - C. Follic acid 400mg per day
   - D. Vitamin B6'B12

2. WHICH OF THE FOLLOWING INVESTIGATION CAN CAUSE MAXIMAUM HARM TO FETUS IF PERFORMED BETWEEN 8-15 WEEKS OF GESTATION
   - A. X ray abdomen
   - B. X ray chest
   - C. Barium enema
   - D. Ct brain –plain

3. LATE DECELERATION OF FETAL HEART RATE IN NST ARISES BECAUSE OF
   - A. Cord compression
   - B. Utero placental in sufficiency
   - C. Fetal head compression
   - D. Normal uterine contraction

4. IN SECOND TRIMESTER USG SEVERE FETAL HYDRONEPHROSIS IS DIAGNOSED AT FETAL RENAL AP DIAMETER OF
   - A. >2mm
   - B. >7mm
   - C. >15mm
   - D. >10mm

5. DECREASED FETAL MOVEMENTS ARE SEEN IN ALL CONDITIONS EXCEPT
   - A. Congenital myopathy
   - B. Spinal muscular atrophy
   - C. Duschenne muscular dystrophy
   - D. Maternal smoking

Answers: Last Page

# Pedigree Charting – A Simple Genetic Tool for Prenatal Diagnosis

by Dr. Dhanlaxmi Shetty, shettydl@tmc.gov.in

**WHAT IS PEDIGREE?**

Pedigree is a short-hand method of describing genetic condition in many generations at a glance. It is an important tool & ethical way for studying inherited familial diseases. Pedigree charting uses family trees and information about affected individuals to:

figure out the genetic basis of a disease or trait from its inheritance pattern( mode of inheritance)

predict the recurrence risk of disease in future offspring in a family (most useful in genetic counseling)

**CONSTRUCTING A FAMILY TREE / PEDIGREE:**


Genetic information is the first and most important step and every clinician can obtain it using Pedigree Chart.

Pedigrees are the preferred method of collecting family history information because a pedigree can be drawn more quickly than the information can be written and allows patterns of disease to emerge as it is drawn.

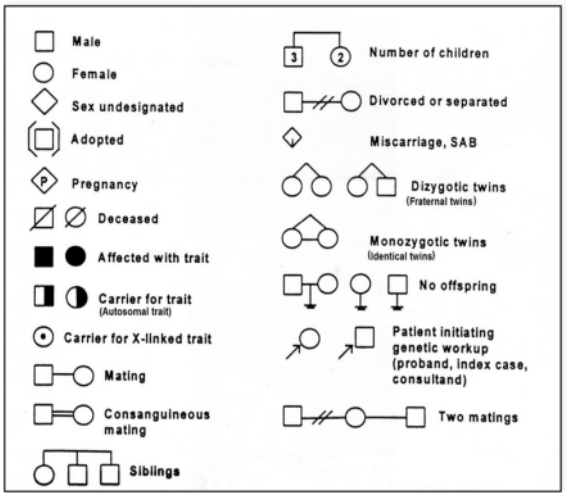
**How to begin taking ' Genetic Family History'?**

- To begin taking a family history, healthcare professionals should ask the patient about his/her health history (present and past) and then ask about siblings and parents. Once done, then ask about aunts, uncles, grandparents, and first cousins.
- To construct a pedigree, it is sufficient to trace a person's ancestry back for about three or four generations.
- It is crucial to distinguish between families with a known genetic or inherited disorder and those with no known disorder. Ask them whether they are carriers of a known genetic disease.
- Note the date when the pedigree was drawn up.
- Mother's maiden name is essential as this is especially significant for X-linked disorders.

**Information to be asked and points to be noted!!**

- General information such as names and birthdates ( check for advanced maternal or paternal age)
- Family's origin or racial/ethnic background
- Health status, including medical conditions (ask if they smoke, if they are overweight, what their diet and exercise habits are) and ages at diagnoses
- Age at death and cause of death of each deceased family member
- Pregnancy outcomes of the patient and genetically-related relatives
- Enquire specifically about high risk factors such as infant deaths, stillbirths and abortions; a serious birth defect / multiple congenital anomalies/ genetic disease ( previous history of Down syndrome…)/ serious developmental delay, or an unexplained abnormality if any.
- Ask whether more than one close relative has the same disease, e.g., mental retardation, learning problems, deafness, blindness, cancer, early heart attacks, or schizophrenia.
- Exposure to teratogens (drugs, X- rays) in pregnancy if any should be recorded.
- Consanguinity, or marriage between close relative should be directly asked about and may be the clue that suggests autosomal recessive inheritance.

**A pedigree represents family members and relationship using standardized symbols (see Pedigree Symbols below)**



- Mistaken or Unacknowledged paternity must be borne in mind, especially in a puzzling situation. Definitive tests of paternity based on DNA can help to resolve the problems.
- Basic details of both the families (paternal as well as maternal) are very essential. Unexpected findings may emerge.

Because the family history continually changes, the pedigree can be updated easily on future visits. Patients should be encouraged to record information and

***Every soul is beautiful and precious; is worthy of dignity and respect, and deserving of peace, joy and love."***
***— Bryant McGill, Voice of Reason***

**How to evaluate a pedigree...**

1) Transmission: Are there affected family members in every generation (vertical pattern) or in only a

single generation (horizontal pattern)?

2) Sex Differences: What is the ratio of affected males to females?

3) Segregation: Is disease/gene being passed through unaffected females? Is there male to male transmission? What % of children are affected (e.g. all of sons but none of daughters)?

**TERMINOLOGIES....**

**Allele -** One or more alternative forms of a gene found at the same (corresponding) locus on homolous chromosomes in an individual &/ or a population.

**Homologous genes** - Identical alleles occupying the same locus on homologous chromosomes.

**Homozygous** - An Individual ( homozygote) who possesses two identical alleles at one particule locus on homologous chromosomes.

**Heterozygous** - An individual ( heterozygote) who possesses two different alleles at one particular locus on homologous chromosomes.

**Dominant -** An allele that is always expressed , both in homozygous and heterozygous conditions.

**Recessive** - An allele that is expressed only when it is homozygous.

**Genotype** - The total genetic constitution ( genome) of an individual, or more specifically, the alleles present at one locus for a particular trait.

**Phenotype** - The appearance( physical, biochemical& physiological) of an individual produced by expression of the genotype under the influence of the environment.

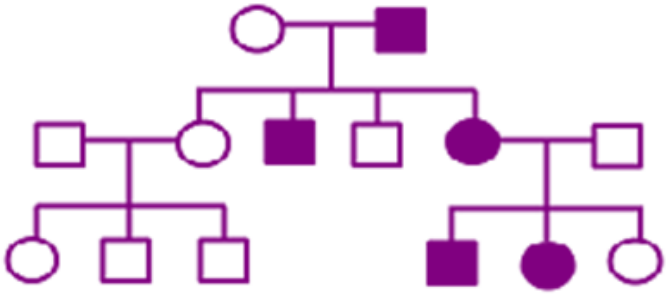
**Let us learn.....**

**AUTOSOMAL DOMINANT INHERITANCE**

- Vertical pattern: multiple generations affected

- Males and females equally likely to be affected

- See male to male transmission



- Each child of an affected individual has a 50% chance to be affected, i.e. 50% risk in every pregnancy

- Unaffected individuals do pass on the gene

- Every affected child has an affected parent

- Example-Polydactyly, Huntington's disease, Achondroplasia (a sketelal disorder causing dwarfism)

**AUTOSOMAL RECESSIVE INHERITANCE**

- Horizontal pattern: single generation affected.

- Males and females equally likely to be affected

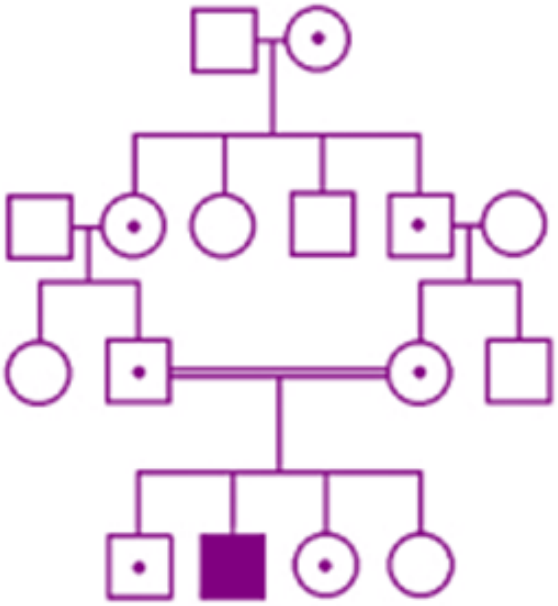
- Increased consanguinity seen

- Parents of affected child are unaffected gene carriers and have a 1 in 4 or 25% recurrence risk

- Unaffected siblings have a 2/3 or 67% chance to be carriers.

-Children of affected individuals are obligate carriers.

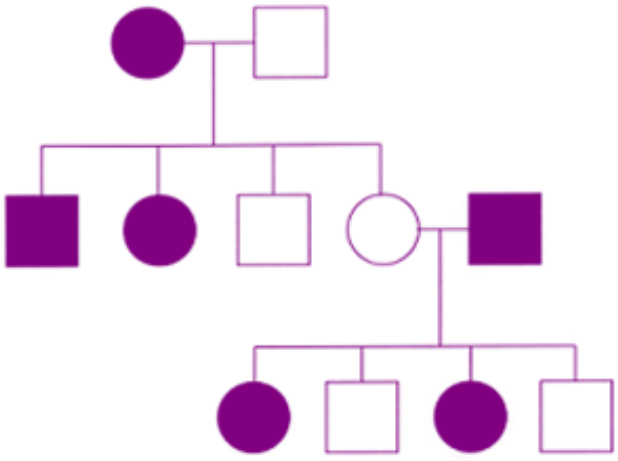
-Example-Cystic fibrosis, Sickle cell anemia, Phenylketonuria (PKU), Tay-Sachs disease



**X-LINKED DOMINANT INHERITANCE**

-For rare conditions, females are about 2 times as likely to be affected than males. May be lethal in males and but variable in females & usually milder,.

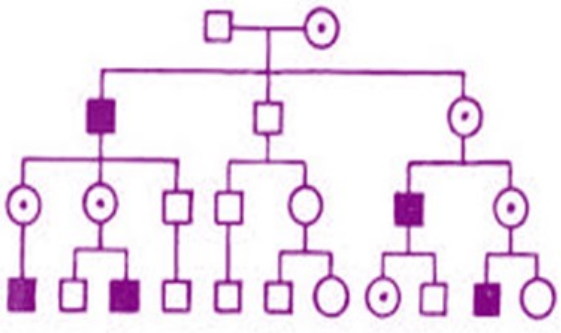
- Affected males pass the gene to all of their daughters, who will be affected, and to none of their sons (No male-to-male transmission)



-Sons and daughters of affected females have 50% chance of being affected (similar to autosomal dominant)

-Example-Incontinentia pigmenti (skin lesions), X-linked rickets (bone lesions)

**X-LINKED RECESSIVE INHERITANCE**



- Males are more often affected than females

- Affected males pass the gene to all of their daughters and none of their sons (No male-to-male transmission)

- Daughters of carrier females have a 50% chance to be unaffected carriers. Sons of carrier females have 50% chance to be affected.

- Affected males in the family are related to each other through carrier females

- For genetically lethal X-linked conditions, 1/3 of isolated cases (i.e. no family history) are new mutations.

- In 2/3 of cases, the mother is an unaffected carrier

- Female gene carriers are usually not affected

- Exceptions: Turner syndrome, skewed X-inactivation, X;autosome translocation carriers

- Example-Colour-blindness, Duchenne Muscular, Dystrophy, Hemophilia

**Basic patterns of inheritance**

| Mendelian Inheritance | Non-Mendelian Inheritance |
|---|---|
| Autosomal dominant | Imprinting |
| Autosomal recessive | Mitochondrial |
| X-linked dominant (very rare) | Multi-factorial |
| X-linked recessive | Sporadic |
| Y – linked | Contiguous gene syndromes |

**but don't forget the complex factors!**

- **Non-penetrance**
- **New mutation**
- **Adult-onset conditions**
- **Consanguinity**
- **Interaction**
- **Sex-limited/sex influenced**
- **Germline mosaicism**
- **Anticipation**
- **Heterogeniety**
- **Pleiotropy**

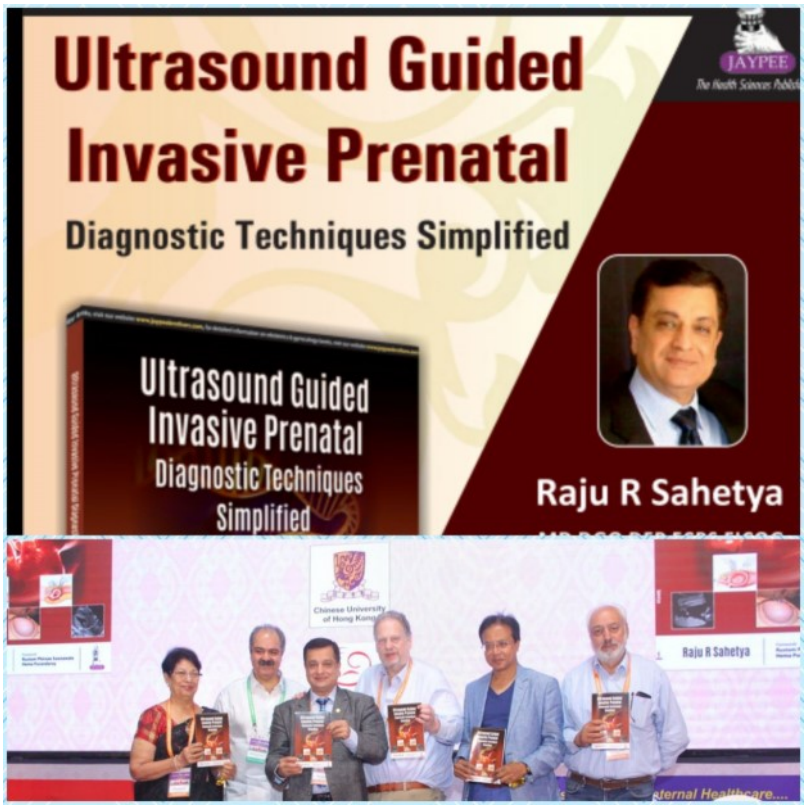
**In brief, complete history taking and pedigree charting is prerequisite for the Laboratory genetic investigations of the Index Case & to calculate and explain the risk of having affected children for future Prenatal Diagnosis.**

**It is ideal to take the help of a qualified Geneticist or Genetic Counselor in case of difficulties.**

## Book release- The comprehensive book - Ultrasound Guided 'Invasive Prenatal Diagnostic Techniques' Simplified…

**Author: Dr Raju R Sahetya**

The grand launch of this book at the 13th Asia Pacific Conference of Fetal Medicine, held in Delhi, 1st to 3rd September 2017, by the hands of none other then Dr Evans Mark from New York, USA, one of the pioneer's in 'Invasive Prenatal Diagnostic Techniques'.




**Dr Evans Mark's remarked** – This book is one of its kind in the field of 'Invasive Prenatal Diagnostic Techniques' a precious possession for all those who practice Fetal Medicine

The comprehensive book - Ultrasound Guided **'Invasive Prenatal Diagnostic Techniques' Simplified…**

The emphasis of this book, however, is not on theory, but on practical techniques, and it is anticipated that by following the methods described here, one will be able to practice interventional ultrasound guided invasive prenatal diagnostic techniques successfully!

Interventional prenatal diagnostic techniques are not commonly taught at graduate or post-graduate training. This is the reason I decided to write a hand book of techniques covering in one book. I have made every effort to share with every reader all the details, description, traps and tricks about the techniques they should know. I hope this book will be a tool to Obstetrician and Ultrasonologist, who can refer this book for learning and training and before performing these techniques.

Answers (November 2017): 1. A, 2. C, 3. B, 4. D, 5. C