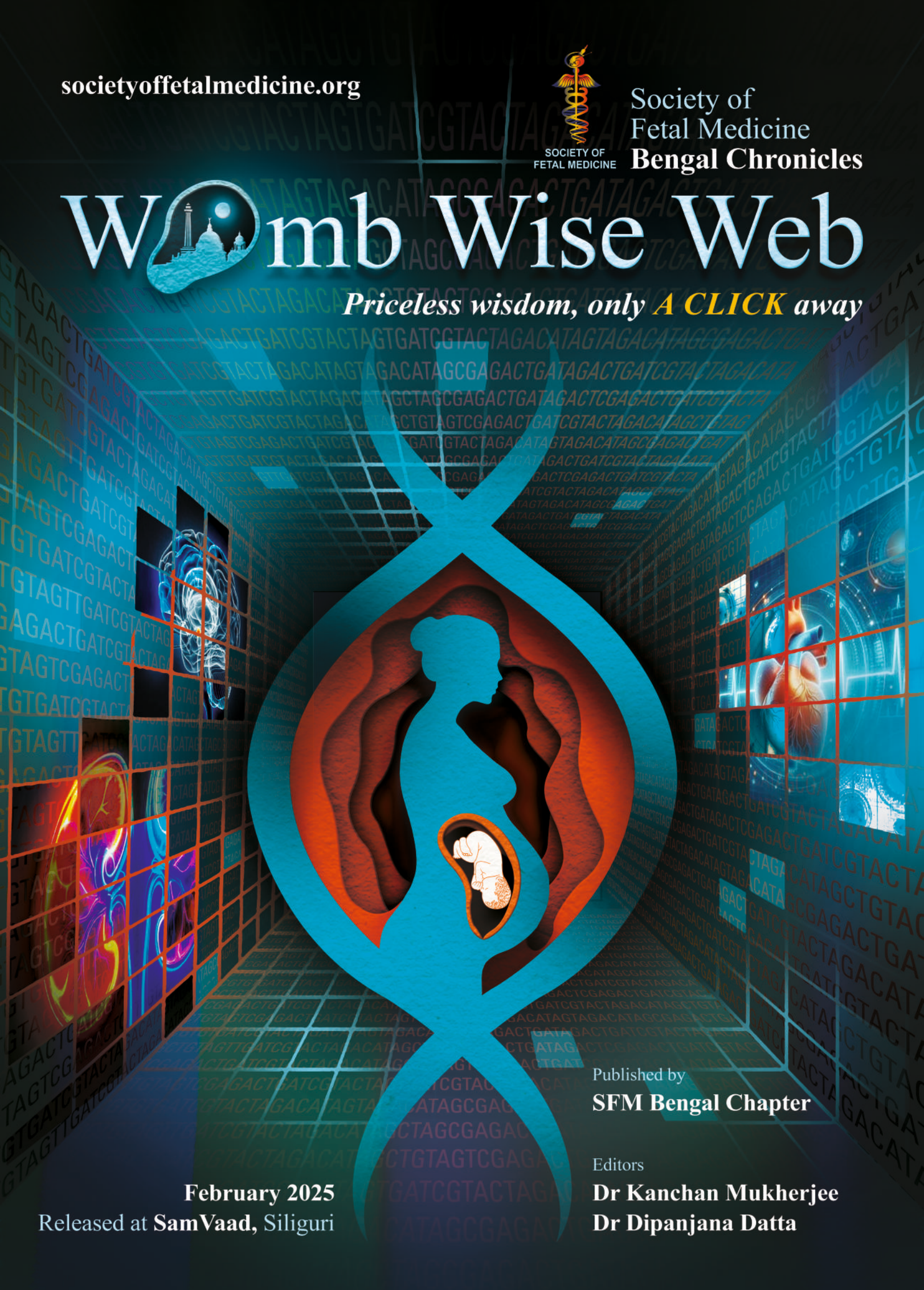
societyoffetalmedicine.org

SOCIETY OF FETAL MEDICINE

Society of
Fetal Medicine
**Bengal Chronicles**

# Womb Wise Web

***Priceless wisdom, only A CLICK away***



Published by
**SFM Bengal Chapter**

Editors
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**February 2025**
Released at **SamVaad,** Siliguri

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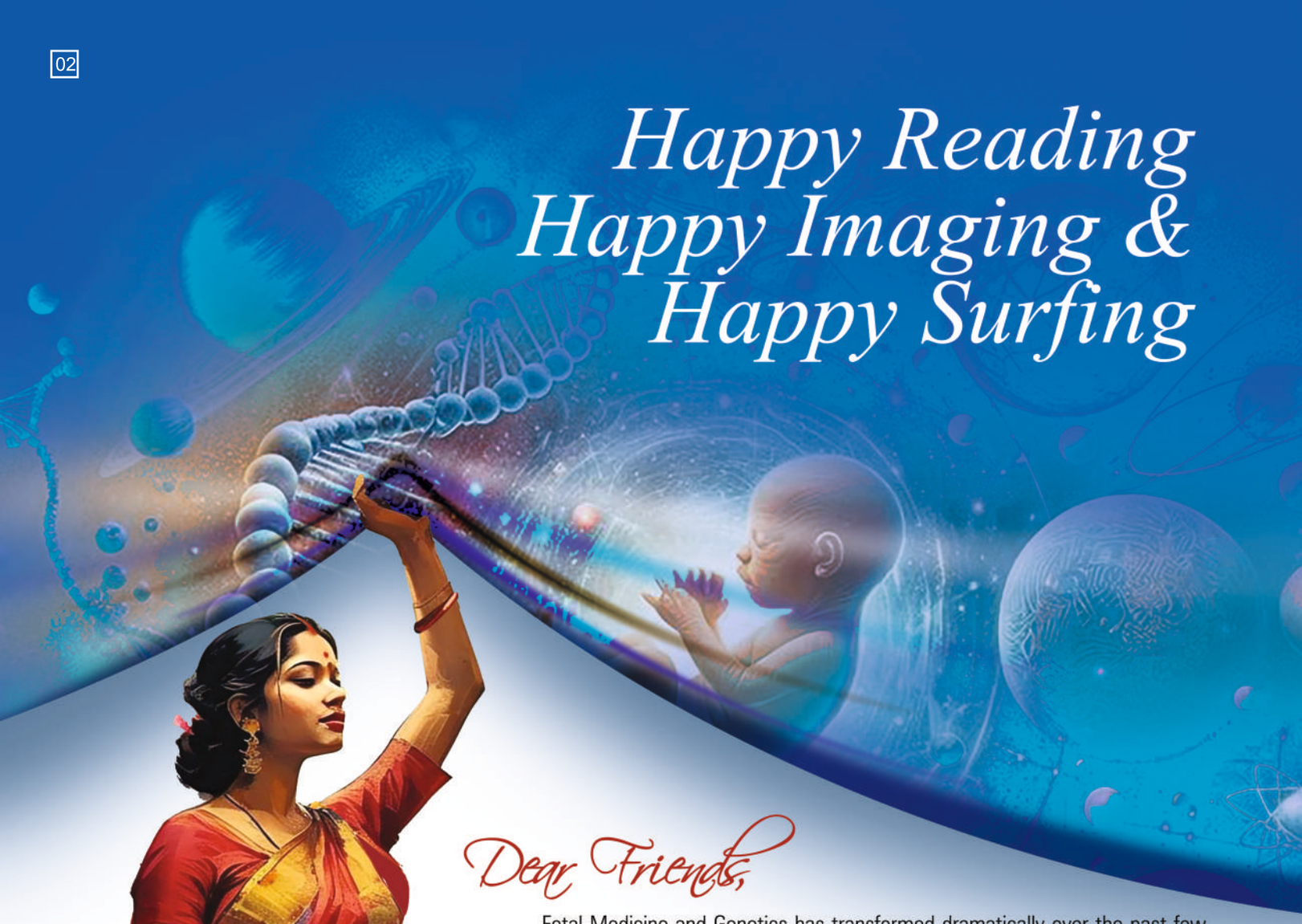
# Happy Reading Happy Imaging & Happy Surfing



## Dear Friends,

Fetal Medicine and Genetics has transformed dramatically over the past few decades and staying updated with the latest research, guidelines and technologies is essential. As busy clinicians, finding accurate and up-to-date information in Fetal Medicine and Genetics is crucial, but with so many websites available, navigating them effectively can be challenging.

Our latest newsletter is here to help!. This innovative newsletter, brainchild of our dear Past President SFM Bengal Chapter, Dr Kanchan Mukherjee, is one of its kind and I can't thank him enough for this. This newsletter is a wonderful compilation of the different websites available to access information on Fetal Medicine and Genetics. The newsletter provides expert insights on using various fetal medicine and genetic websites effectively – helping you access the latest research, guidelines and essential tools with ease.

I am extremely thankful to Dr Dipanjana Datta for helping us get articles on various genetic websites and grateful to all the contributors for sparing their valuable time in providing their articles. A special thanks to our Mentor Emeritus Dr Ashok Khurana and Dr Mohit Shah, President SFM for allowing us to release this newsletter at the prestigious SamVaad. I must acknowledge team Rueda for their creative ideas in designing this newsletter for you.

So friends, stay informed, stay updated and make the most of the trusted Fetal Medicine and Genetics resources.

Happy reading, Happy Imaging and Happy Surfing!

Long live SFM

Jai Hind

**Seetha Ramamurthy Pal**
President SFM, Bengal Chapter

# SFM Digital Ecosystem: Empowering Fetal Medicine Practitioners Worldwide


**Mr Faisal Khan**
Editorial Administrator
(Journal of Fetal Medicine) &
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The Society of Fetal Medicine (SFM) is a global forum of Indian origin dedicated to offering every fetus an optimal outcome. Recognizing the critical importance of continuous education and accessible resources for fetal medicine practitioners, SFM has developed a comprehensive suite of online platforms. These include an informative website, an engaging YouTube channel, and active social media presences on X (formerly Twitter), Instagram, and Facebook. Each platform is meticulously designed to support healthcare professionals in staying abreast of the latest advancements, guidelines, and best practices in fetal medicine.

## The Website

SFM's official website serves as a centralized hub, offering a plethora of resources tailored to meet the diverse needs of fetal medicine practitioners. The Practice Guidelines, such as those for the Second Trimester Anomalies Scan, Invasive Procedures, and Fetal Therapy including intrauterine blood transfusion, fetal reduction, fetal shunt, laser ablation, etc. are instrumental in enhancing diagnostic accuracy, improving overall standards, and ensuring medico-legal safety in practice. The site is replete with educational materials, including newsletters and e-books, covering various topics in fetal medicine. These resources are designed to keep practitioners informed about the latest research findings, technological advancements, and clinical updates. Regular engagement with these materials fosters a culture of continuous learning and professional development. Additionally, the website maintains an archive of past events, including webinars and conferences, allowing practitioners to revisit and learn from previous sessions that cover a broad spectrum of topics pertinent to their daily work, as well as upcoming events featuring cutting-edge advancements in fetal medicine, hands-on workshops led by global experts, and interactive panel discussions on emerging challenges in prenatal diagnostics and therapy. These forthcoming events provide a platform for continuous professional development, offering exclusive insights into novel imaging techniques, fetal interventions, and evolving clinical guidelines, ensuring that healthcare providers remain at the forefront of medical excellence in fetal care.

## The YouTube Channel

Complementing the resources on the website, SFM's YouTube channel offers a rich repository of video contents designed to cater to audio-visual learners. The channel features recordings of webinars on diverse topics providing in-depth discussions and insights into various feto-maternal conditions. These are invaluable for practitioners aiming to broaden their understanding of complex issues. Clinical case discussions present real-life scenarios with practical insights into diagnostic challenges and management strategies, bridging the gap between theory and practice and enhancing clinical decision-making skills. The channel also regularly uploads discussions with leading experts in the field, providing viewers with access to a wealth of experience and knowledge.

## X (Formerly Twitter) Handle

SFM maintains an active presence on X under the handle @Socfetmed. This platform serves as a dynamic space for real-time updates, professional discourse, and community engagement. It facilitates interactive discussions among practitioners, allowing for the exchange of ideas, experiences, and best practices, thereby fostering a sense of community and collective growth. Timely dissemination of information about upcoming events, recent publications, and organizational news keeps SFM members informed and engaged. The curated content supports practitioners in staying informed and enhancing their clinical practice.

## Instagram and Facebook

Beyond X, SFM actively engages with the community on Instagram and Facebook (both@societyoffetalmedicine). These platforms are utilized to share visual content, including infographics, event highli-ghts, and educational materials, catering to a broad audience. Followers can stay updated on the latest developments in fetal medicine, view event announc-ements, and access a variety of educational resources. The interactive nature of these platforms encourages community engagement, allowing practitioners to connect, share insights, and discuss pertinent topics in fetal medicine.

## Membership Benefits

Becoming a member of SFM opens doors to a global community of professionals committed to fetal health. Membership at a nominal fee provides privileged access to specialized resources, including advanced practice guidelines, detailed case studies, and proprietary research findings.

### Salient features

- Comprehensive Online Resources
- Evidence-Based Practice Guidelines
- Rich Educational Content
- Free-to-Publish Open-Access Journal
- Active Professional Community
- Global Reach and Collaborations

Members also benefit from networking opportunities, connecting with peers in fetal medicine to facilitate collaborative projects. Engaging in the academic programs organized by SFM ensures that practitioners remain at the forefront of advancements in fetal medicine. Additionally, SFM continuously collaborates with international organizations to enhance its global reach, providing members with access to additional educational materials, research opportunities, and global best practices.

### SFM Official Journal

The Journal of Fetal Medicine is the official peer-reviewed, open-access journal of the Society of Fetal Medicine, dedicated to advancing research and clinical practices in fetal medicine. Covering original research, review articles, case reports, and practice guidelines, it serves as a vital platform for specialists worldwide. Uniquely, it is a free-to- read and publish journal with zero article processing charges, as all costs are fully borne by SFM. This ensures that researchers and practitioners can freely share and access the latest developments, technological advancements, and clinical insights without financial barriers, fostering continuous learning.

The Society of Fetal Medicine's commitment to advancing fetal health is evident through its extensive online presence. By leveraging the tools and information available on their official website, YouTube channel and social media platforms, practitioners can stay abreast of the latest developments, refine their clinical skills and contribute to the overarching goal of ensuring optimal outcomes for every fetus.

## Dive Deeper

SOCIETY OF FETAL MEDICINE

# ISUOG – Disseminating Highest Quality Education Online

**Dr Shikha Marwah**
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Kolkata

## Introduction

The International Society of Obstetrics and Gynaecology (ISUOG) was founded in 1991 by Prof Stuart Campbell. It is a professional membership association and charity registered in England and Wales. ISUOG represents and supports professionals using ultrasonography in obstetric and gynaecological practice across the world. ISUOG's long-term vision is that every woman in the world has access to ultrasound, that every scan provider is competent and that the diagnosis of obstetric and gynaecologic conditions is effective so that women's health outcomes improve.

## Content and practical application

ISUOG provides various educational opportunities to its members.

### Basic training:

It focusses on standardizing basic approach to OB/GYN ultrasound. Carefully selected topics in obstetric and gynaecological ultrasound are dealt with in a comprehensive way, developed by experts in the field. ISUOG basic training course is useful for medical professionals who are new to ultrasound, to acquire knowledge and skills for performing basic ultrasound and to understand as to when to refer patients to a higher center.

### Practice guidelines:

ISUOG Practice Guidelines contain research and recommendations for obstetrics and gynaecology professionals to assist in maintaining the highest possible clinical practice, to improve and standardise medical care for our ultrasound in OBGYN speciality. For eg, Updated ISUOG Practice Guidelines: role of ultrasound in twin pregnancy addresses the role of ultrasound in the care of uncomplicated and complicated twin pregnancies.

### ISUOG patient information leaflets:

These are written and reviewed by medical professionals for patients who need quick and reliable information. The series covers a range of topics in obstetrics and gynaecology to help patients and their family members make more informed choices.

### Ultrasound in Obstetrics & Gynaecology:

UOG is the official, monthly, peer-reviewed journal of the ISUOG, featuring the latest clinically relevant research, including guidelines, consensus statements, expert commentaries, original articles, and systematic reviews. Each month at least one article is selected for free access.

### VISUOG:

Visual encyclopedia, provides up-to-date information on the clinical use of ultrasound in obstetrics and gynaecology. It is exclusively available to ISUOG members. However, some of the contents are often freely accessible.

### Calculators:

Various calculators like fetal biometry are available for online reference, however, with membership.

## Dive Deeper

INTERNATIONAL SOCIETY OF OBSTETRICS AND GYNAECOLOGY (ISUOG)

## ISUOG

ISUOG is an important society of fetal medicine speciality. It provides educational opportunities to keep oneself updated in the ever-developing and evolving field of ultrasound in obstetrics and gynaecology.

# Fetal Medicine Foundation (FMF): The Guiding Star

**Dr Arkajyoti Mukherjee**
Fetal Medicine Consultant
Apollo Multispeciality Hospital
Kolkata

For improvement of pregnancy and fetal care FMF is working for more than 3 decades. The online resources through their website aims to set a standard in fetal medicine practice across the countries. From educational materials to online prediction and assessment algorithms, all are just a click away and absolutely free of cost. For both the busy practitioners and the beginners it provides short yet precise content through resumable modules encompassing almost all the common aspects of fetal imaging and its application in clinical practice. The site contains different segments.

## EDUCATION

1. **11-13 weeks scan-** Imaging guidelines, aneuploidy screening and abnormalities which can be picked up through this scan are well explained.
2. **Fetal abnormalities-** System wise insight on fetal anatomy scan, the common abnormalities, associated genetics and management plan are there.
3. **Fetal echocardiography-** Specific materials on normal and abnormal fetal heart.
4. **Preeclampsia screening-** History, doppler and biochemistry-based prediction of PE and FGR.
5. **Cervical assessment-** When and how to measure along with prediction of preterm birth.
6. **Placenta accreta spectrum-** The sonographic features and implications.
7. **Doppler ultrasound-** How to properly assess and interpret MCA, Umbilical, Ductus Venosus and Uterine artery dopplers.

## CERTIFICATIONS AND LICENSE

Certificate of competence for Nuchal Translucency, Nasal Bone, Tricuspid, Ductus Venosus, Uterine artery doppler, Cervical length and Doppler imaging are issued upon completion of online modules and submitting proper images following their structured guidelines. For fetal abnormalities one has to complete online modules, submit logbook of images and appear in practical examination at an FMF approved centre. Certificate for Fetal Echocardiography and invasive procedure are reserved for FMF fellows. On achievement of certificate of competence for 11-13 weeks scan FMF also provides annually renewable license for accessing their free available First Trimester aneuploidy risk calculation algorithm software and annual performance audit.

## CALCULATORS

Various freely accessible **prediction** (Aneuploidy, SGA, FGR, Preeclampsia, Preterm, GDM, Macrosomia), **management** (SGA, Fetal Anaemia) and **assessment** (NT, Growth and Dopplers) calculators are available, which are quite essential for our day to day practice. If someone does not have reporting software these calculators help to generate a standard and meaningful report. The obstetricians can also interpret a 'not so standard' report in their clinical practice with the help of such tools and make appropriate management plan.

## OTHER INFORMATION

Information regarding upcoming courses and congress, details of FMF fellowship and diploma programme are also provided and updated there. Moreover there are loads of research publications, common and rare case reports, trials available which are essential for everyone's academic upliftment.

## Dive Deer

FETAL MEDICINE FOUNDATION

## Salient features

- Tailor made study materials
- Certificate of competence
- License and free software
- Calculators- Fetal anemia, FTS, Preeclampsia, Growth and Doppler etc
- Publications
- Courses and conferences

# Fetal Medicine Barcelona – Offering Advanced Education in Maternal-Fetal Care

**Dr Pooja Vaziraani**
MD Obgyn, DNB, FCPC, DGO
Fellowship in Fetal Medicine (Mediscan Chennai)
Expert in Imaging in Maternal-Fetal medicine UBCN, Spain)
International Master in Maternal Fetal medicine (UBCN, Spain)
Postgraduate in Fetal Cardiology (UBCN, Spain)
Fetal Medicine Consultant

Fetal Medicine Barcelona website offers information regarding our portfolio of training programs and projects in the field of obstetrics and gynecology and fetal medicine. Additionally, it offers learning resources that deliver a premium experience to doctors through tools such as protocols, calculators, e-learning, apps, and free masterclasses and workshops.

Target: Fetal medicine, gynecology and obstetricians specialists including clinicians, and radiologists.

*https://fetalmedicinebarcelona.org/en/*

## PROGRAMS

### MATERNAL FETAL MEDICINE

**Hybrid Learning (Paid courses)**

- International Master in Maternal-fetal Medicine
- Training Course in Imaging in Maternal-Fetal Medicine

**Online Training: (Free)**

- Free International Interactive Masterclass: Update in Prematurity

### MATERNAL MEDICINE - HIGH-RISK OBSTETRICS

**Hybrid Learning**

- Postgraduate Course in High-Risk Obstetrics

### FETAL MEDICINE

**Hybrid Learning**

- Postgraduate Course in Fetal Cardiology
- Postgraduate Course in Fetal Neurology
- Postgraduate Course in Fetal Medicine – India

**Online training (paid online lectures and MCQS with certification at the end of the MCQS)**

- Advanced Fetal Echocardiography
- Advanced Fetal Neurosonography
- Fetal Pathology: Perinatal Diagnosis and Management
- First Trimester Prenatal Screening and Diagnosis
- Infections in Fetal-Maternal Medicine
- Placental Disease, Intrauterine Growth Restriction and Preeclampsia

### INTERACTIVE COURSES

- Interactive course in fetal anatomic ultrasound 2.0 (Paid course)

### EXTERNAL COLLABORATIONS (paid courses)

- Fellowship Cardiology
- Fellowship in Maternal fetal medicine

### FREE TOOLS:

- Protocols
- Calculators

### PROTOCOLS IN MATERNAL-FETAL MEDICINE (MFM)

*https://fetalmedicinebarcelona.org/protocolos/*

The medical protocols encompass information intended as guidance for diagnosis and the treatment of specific pathologies. However, it is evident that these guidelines necessitate adaptation to the specific clinical situation and context, taking into consideration the doctor's clinical judgement.

These protocols serve as professional guidelines intended for specialist doctors. Correct interpretation requires proper training, and the concepts expressed within should not be applied directly to patients without considering the specific details of each individual case.

**Below is the list of protocols available in English that will benefit everyone in the field of MFM**

- Alloimmunization in pregnancy
- Amniocentesis
- Anaemia during pregnancy and postpartum period
- Antenatal management in pregnant women with previous cesarean section
- Antepartum Fetal Surveillance
- Care for preterm birth in singleton pregnancies
- Cordocentesis
- Chorioamnionitis
- Cervical Cerclage
- Corticosteroids
- Decrease of Fetal Movements (DFM)
- Doppler
- Ectopic Pregnancy
- Fetal arrhythmias
- Fetal echocardiography
- Fetal growth defects
- Fetal growth disorders
- Fetal screening protocol
- First trimester pregnancy loss
- Genetic studies in Fetal Samples
- Gestational Diabetes

## Salient features

- Fetal medicine topics
- Patient-friendly resources
- Educational materials for healthcare professionals
- Easy to use, up-to-date calculators designed using published data
- Protocols based on evidence-based medicine followed in their dept of obstetrics and fetal medicine

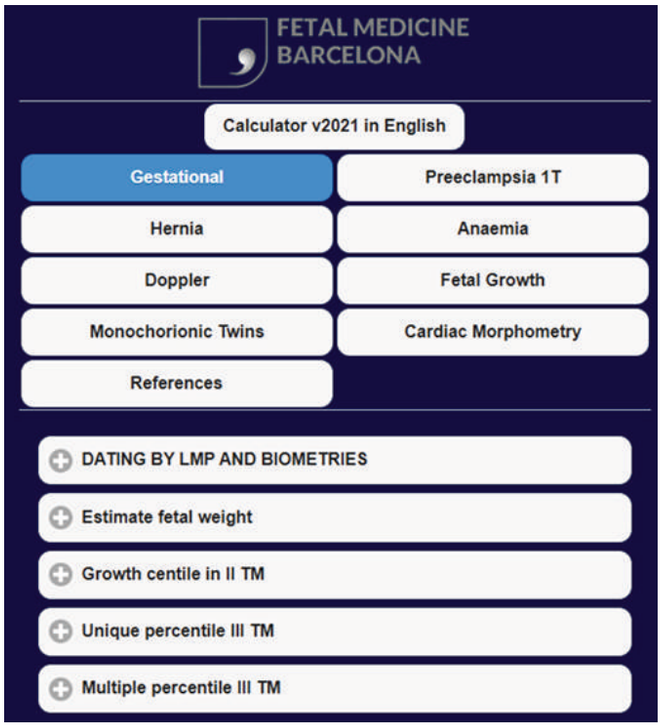
- Hepatitis and Pregnancy
- Hyperemesis Gravidarum
- Hypertension and pregnancy
- Induction of Labour and Cervical Ripening
- Intrahepatic Cholestasis of Pregnancy
- Intrapartum and puerperal fever
- Intrapartum Assessment of Fetal Well-being
- Listeria and Pregnancy
- Macrosomia
- Management of hydatidiform mole
- Management of Multiple Pregnancy
- Management of patients at risk of preterm delivery
- Monochorionic twin Pregnancy: Selective Fetal Growth Restriction (sFGR)
- Monochorionic Twin Pregnancy: Twin-To-Twin Transfusion Syndrome (TTTS)
- NGS Fetal Neurosonography Clinical Guide
- Non-Immune Hydrops Fetalis
- Oligohydramnios in singleton gestation
- Placental anomalies and third trimester bleeding
- Placental Haematomas. Placental Abruption
- Polyhydramnios in Singleton Gestation
- Postpartum haemorrhage
- Pregnancy of Unknown Location – PUL
- Prenatal screening for chromosomal defects LBQ
- Preterm Labour
- Preterm prelabour rupture of membranes at term and preterm
- Shoulder Dystocia
- Second Trimester Pregnancy Loss and Fetal Death
- Sepsis and Septic Shock in Pregnancy and Puerperium
- Systemic Lupus Erythematosus
- Torch infection in pregnancy
- Urinary Tract infection and pregnancy
- Venous Thromboembolism Prophylaxis in Pregnancy and Puerperium

## CALCULATORS

*https://fetalmedicinebarcelona.org/calc/index.html*

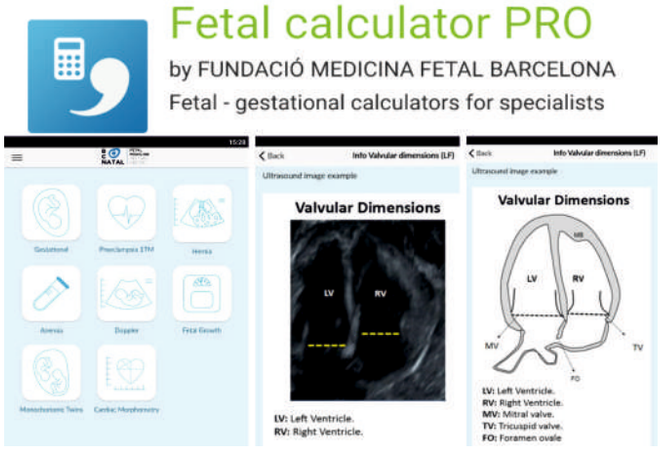
The BCNatal calculators based on multiple research studies are freely available on Internet providing different calculators in the following areas:

- Gestational
- Doppler
- Fetal Anemia
- Preeclampsia
- Fetal Growth
- Congenital Diaphragmatic Hernia




For offline use, a calculator app is also available for iPhone and iPad.

*https://apps.apple.com/in/app/fetal-bcn-calculator/id*



# Perinatology.com: Free Web Resources for Fetal Medicine Practitioners

**Dr Urvashi Chhikara**
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New Delhi

Perinatology.com is a freely accessible website, whose primary focus is to provide elaborate information to the Fetal medicine specialists, obstetricians, practitioners and patients regarding various aspects of pregnancy and conditions associated with it.

## Introduction

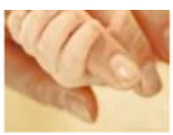
Perinatology.com is maintained by Focus Information Technology, a private company engaged in the development of ultrasound reporting systems and related websites, as a resource for perinatologists (maternal-fetal medicine specialists), obstetricians, ultrasonographers and genetic counsellors.

## Content Highlights

Home page of the website is divided into 3 main sections - various calculators, nomograms, z-scores and charts; education and certification; and third section including general information on drugs, infections in pregnancy, genetics, various protocols and patient information leaflets.

## Practical Applications

Perinatology.com is a robust and elaborate website, providing not just an easy access to the FM specialists and care providers, but also acts as a pregnancy guide for the patients. The various services provided by the website are listed below.



perinatology.com

Search Perinatology for Go

Calculators
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Glossary
Genetics
Instructional
Intrapartum Fetal Monitoring
OBPharm
Translate this website

Patient Information
Perinatologists
Protocols
Questions
Reference Ranges
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MOST FREQUENTLY VISTED

- **Pregnancy Due Date Calculator**
- Due Date from Ultrasound Report
- Gestational Age On/ Fetal Development
- Estimation of Fetal Weight
- Maternal Age at Delivery
- Midtrimester Risk for Chromosome Abnormalities
- Caloric Requirements and Initial Insulin
- **Expected Peak Velocity of Systolic Blood Flow through MCA**
- **Fetal Biometry 3.0**
- Reference Ranges
- Bishop Score
- Fetal Development
- Beta hCG Doubling Time
- Drugs in Pregnancy

***Fig 1- Home page of perinatology.com***

Pregnancy dating is available from last menstrual period, from IVF dates (date of embryo transfer), from ultrasound, and it also provides the expected date of delivery, period of gestation on a certain given date and also gives a guide for important antenatal scans and blood tests at various gestations of the pregnancy.[1]

perinatology.com
Pregnancy Due Date and Gestational Age Calculator

Home > Calculators >Pregnancy Due Date Calculator

Calculate estimated due date (EDD) and gestational age based on :

- Conception date (date of ovulation, egg retrieval, or insemination)
- Date of 3-day embryo transfer
- Date of 5-day embryo (blast) transfer
- Due date by sonogram (reverse due date calculator)
- First day of last menstrual period

month date year

Calculate

See Also
Calculate Due Date From Ultrasound Report
Calculate Gestational Age from Ultrasound Measurements
Calculate Gestational Age on a Given date.

***Fig 2- Pregnancy dating calculators, calculator for gestational age on certain date***

The website also helps in guiding the early pregnancy by providing beta-HCG doubling time calculator. Provides reference range for beta-HCG (using the Roche Cobas analyzer) during the first trimester of pregnancy.[2]

It has got normal trimester wise reference range for lab values in pregnancy, along with their references (with the PMID number, to free access the paper) and also discusses the causes for abnormal values which make it a complete guide for the pregnancy.[3]

It provides patient information regarding fetal development according to the weeks of gestation. Also, various medications and drug exposure in pregnancy and its safety with use is provided.

Calculates the estimated maternal age at the expected date of her delivery, and whether she is advanced maternal age or not.

It acts as a guide for the expecting mother as it will estimate the BMI, recommended weight gain and energy requirement for a singleton or twin pregnancy.

Also gives a guide for Insulin regimen using multiple daily injections of rapid acting, regular Insulin and NPH Insulin, additional diabetes related calculators- estimate initial continuous subcutaneous insulin Infusion (CSII), Insulin pump settings, average blood sugar levels based on percentage of HbA1c levels.[4] Also provides free patient educational material on food guides, weight gain grid.

The website provides a guide for first trimester screening as it calculates the estimated gestational age according to the CRL. The calculator will also give the percentile for a measured NT if entered for CRL between 45-84 mm, which guides in the aneuploidy screening in the first trimester.[5]

It also provides calculation of age-adjusted ultrasound risk assessment based on mid-trimester genetic sonogram. It also provides calculators for nasal bone length for various ethnic groups, as we understand that normal nasal bone length during the second trimester appear to vary according to race and ethnic background , the use of single fixed cut-off value may be unsuitable for predicting Down syndrome in all populations.

Provides calculators for fetal biometry, based on stated gestational age and measured value for head circumference, thoracic circumference, abdominal circumference, estimated fetal weight, long bones, fetal Dopplers according to various nomograms available and also gives the z score of that parameter for its interpretation. Also provides threshold values for MCA-PSV, FL:AC ratio, CPR, facial angles (inferior facial angle, jaw angle), atrial diameter of lateral ventricles.[6]

Perinatology.com provides guide for fetal echocardiography, provides measurements of various cardiac biometry and z score according to the gestational age. Also M mode indices and pulse wave Doppler indices with the Z score is provided.7 Cardio-thoracic ratio can be calculated for the gestation, especially useful if any cardio-thoracic or skeletal abnormality is suspected. Provides parameters for skeletal survey- HC, all long bones, foot, AC- with percentage and $^{Z}$ score. Also provides important ratios- femur to foot ratio, FL:AC ratio, thoracic to abdominal circumference- which helps to differentiate lethal from non-lethal types of skeletal dysplasia. According to the Z scores it also provides interpretation of the findings and probable causes if abnormality is suspected, for better understanding of the clinician.[8]

It also acts as a guide for monitoring and management of Rh iso-immunised pregnancy. Calculators are available for calculating observed MCA peak systolic velocity as a function of the gestational age. If MCA-PSV is >1.5 MOMs, it is suggestive of fetal anaemia. It also provides the right technique of acquiring MCA Dopplers as it is subject to variation if inaccurate technique is used.[9]

Once fetal anaemia is diagnosed and intra-uterine transfusion is planned, there are calculators for intra-vascular fetal transfusion according to the pre- and post-transfusion and donor haematocrit. Also dose of Pancuronium according to the estimated fetal weight, required for muscular paralysis during the procedure can also be calculated. The technique and methodology for the procedure is provided too.[10]

In cases of suspected lung lesions, it provides calculators for CVR ratio (CPAM volume ratio) and LHR (lung head ratio) in cases of congenital diaphragmatic hernia. It guides regarding the methodology for calculating the CVR or LHR ratio, and also mentions the various prognostic indicators- if CVR is >1.6 or CPAM with dominant cyst, there are high chances of fetal hydrops and 2-3 times per week monitoring is required. Similarly in cases of right or left sided diaphragmatic hernia, based on observed to expected LHR ratio, neonatal survival can be predicted.

- CVR = (Length X Height X Width X 0.52 )/Head Circumference

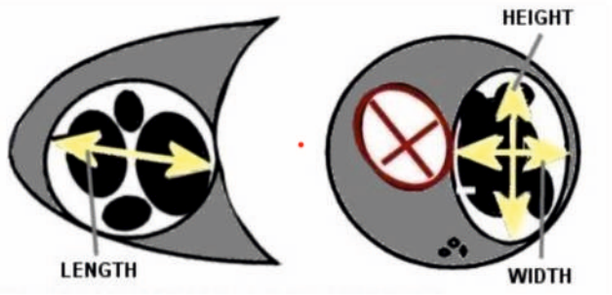


***Fig 3- Methodology for calculating CVR ratio (for congenital pulmonary airway malformation)***

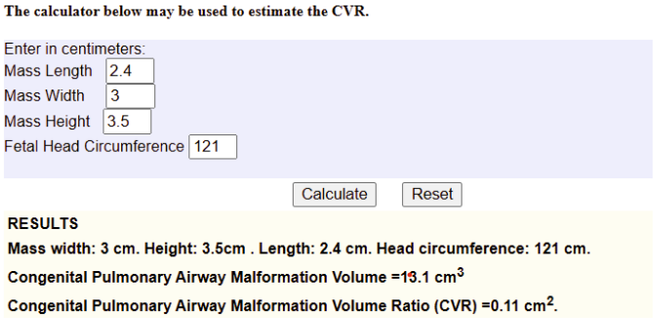


***Fig 4- Calculation of CVR ratio***

Extremely preterm birth outcome tool, by NIH- available for birth upto 25 weeks- information on neonatal survival and morbidity- profound, moderate-severe neuro-developmental impairment, cognitive developmental delays, blindness, deafness, cerebral palsy is provided.

Perinatology.com has been provided with a genetic counsellor toolbox, where it provides information of birth defects and other health conditions and genetic references for those, also information on various genetic tests and carrier screening is available. List on various fetal therapy centers in the United States is provided.

The website also provides access to the recently published abstracts and research papers in international Journals- AJOG (American Journal of Obstetrics and Gynecology), American Journal of Perinatology, British medical Journal, Prenatal Diagnosis, fetal diagnosis and therapy, Lancet, NEJM etc. It also provides access to information for various courses, certifications and any upcoming events, under American Board of Obstetrics and Gynecology, Fetal Medicine Barcelona, Fetal Medicine Foundation.

## REFERENCES


1. Savitz DA, Terry JW Jr, Dole N, Thorp JM Jr, Siega-Riz AM, Herring AH.Comparison of pregnancy dating by last menstrual period, ultrasound scanning, and their combination. Am J Obstet Gynecol. 2002 Dec;187(6):1660-6. PMID:12501080
2. Doubilet PM, Benson CB.Further evidence against the reliability of the human chorionic gonadotropin discriminatory level.

   J Ultrasound Med. 2011 Dec;30(12):1637-42. PMID: 22123998
3. Abbassi-Ghanavati M, Greer LG, Cunningham FG. Pregnancy and laboratory studies: a reference table for clinicians. Obstet Gynecol. 2009 Dec;114(6):1326-31. PMID:19935037
4. Jovanovic L. Role of diet and insulin treatment of diabetes in pregnancy. Clin Obstet Gynecol 2000; 43(1): 46-55 PMID: 10694987
5. age (5-18 weeks) with high-resolution real-time US. Radiology. 1992 Feb;182(2):501-5.PMID: 1732970
6. Hadlock FP, et al., Estimating fetal age:Computer-assisted analysis of multiple fetal growth parameters. Radiology 1984; 152:497-5011. PMID: 6739822
7. Gagnon C, Bigras JL, Fouron JC, Dallaire F. Reference Values and Z Scores for Pulsed-Wave Doppler and M-Mode Measurements in Fetal Echocardiography. J Am Soc Echocardiogr. 2016 May;29(5):448-460.e9. doi: 10.1016/j.echo.2016.01.002. Epub 2016 Mar 9. Erratum in: J Am Soc Echocardiogr. 2019 Jan;32(1):169-170. PMID:26971082
8. Savarirayan R, Rossiter JP, Hoover-Fong JE, Irving M, Bompadre V, Goldberg MJ, Bober MB, Cho TJ, Kamps SE, Mackenzie WG, Raggio C, Spencer SS, White KK; Skeletal Dysplasia Management Consortium. Best practice guidelines regarding prenatal evaluation and delivery of patients with skeletal dysplasia. Am J Obstet Gynecol. 2018 Dec;219(6):545-562. doi: 10.1016/j.ajog.2018.07.017. Epub 2018 Jul 23. PMID: 30048634.
9. Society for Maternal-Fetal Medicine (SMFM). Mari G, Norton ME, Stone J, Berghella V, Sciscione AC, Tate D, Schenone MH. Society for Maternal-Fetal Medicine (SMFM) Clinical Guideline #8: the fetus at risk for anemia--diagnosis and management. Am J Obstet Gynecol. 2015 Jun;212(6):697-710. PMID:25824811
10. Socol ML, MacGregor SN, Pielet BW, Tamura RK, Sabbagha RE. Percutaneous umbilical transfusion in severe rhesus isoimmunization: resolution of fetal hydrops. Am J Obstet Gynecol. 1987 Dec;157(6):1369-75. PMID: 3122574; UI: 88103789


**Dive Deeper**

PERINATOLOGY.COM

# Unlock Digital Freebies Bundled with Your Machine: Boost Your Knowledge

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When we buy any ultrasound machine of our choice, we must think that it comes with great resources for education as a company policy on medical education for the users. I've found some fantastic resources provided by the manufacturers as discussed in this write up below.

## Voluson™ Club: A Hub for Fetal Medicine Experts

The Voluson™ Club is an exclusive online community designed specifically for users of GE Healthcare's Voluson ultrasound systems. This platform is a treasure trove of resources aimed at enhancing the clinical practice, research, and education of fetal medicine practitioners. Whether you are a seasoned professional or just starting in the field, the Voluson™ Club offers a wealth of information and tools to support your work.

**Website: *https://www.volusonclub.net***

## Educational Resources

One of the standouts features of the Voluson™ Club is its extensive Learning Center. Here, members can access a variety of educational materials that cater to various aspects of fetal medicine. These resources include quick cards, guides, white papers, and publications that provide in-depth knowledge and practical tips.

For example, the "First Trimester Screening" guide is an invaluable resource for practitioners. It offers detailed protocols and measurement techniques to ensure accurate and consistent results during the first trimester of pregnancy. This guide helps practitioners understand the nuances of early fetal development and the importance of precise measurements in screening for potential anomalies.

## Clinical Specialties

The Voluson™ Club website is organized into dedicated sections for various clinical specialties, making it easy for practitioners to find relevant information. These specialties include fetal heart, first trimester, gynecology, and assisted reproductive technology. Each section is packed with specialized content designed to support practitioners in these areas.

**Fetal Heart:** This section includes case studies and image libraries that demonstrate both normal and abnormal findings. These resources are crucial for diagnosing congenital heart defects and other cardiac anomalies. By studying these cases, practitioners can improve their diagnostic skills and stay updated with the latest advancements in fetal cardiology.

**First Trimester:** The first trimester is a critical period in fetal development, and this section provides comprehensive information on screening and diagnostic techniques. It includes guidelines on nuchal translucency measurement, nasal bone assessment, and other key markers that are essential for early detection of chromosomal abnormalities.

**Gynecology:** This section offers resources on various gynecological conditions and their ultrasound findings. It covers topics such as ovarian cysts, uterine fibroids, and endometriosis, providing practitioners with the knowledge needed to accurately diagnose and manage these conditions.

**Assisted Reproductive Technology (ART):** The ART section focuses on the role of ultrasound in assisted reproduction. It includes information on follicular monitoring, embryo transfer, and other procedures that are integral to successful ART outcomes. Practitioners can learn about the latest techniques and technologies that enhance the effectiveness of ART treatments.

## Product Information

The Voluson™ Club also provides detailed information about Voluson products, including the latest innovations and technologies. This section is particularly useful for practitioners who want to stay updated with the advancements in ultrasound technology and how they can be applied in clinical practice.

Voluson E10 System: One of the highlights is the Voluson E10 system, known for its advanced imaging capabilities and user-friendly interface. This system is designed to improve diagnostic accuracy and efficiency, making it a valuable tool for fetal medicine practitioners. The Voluson E10 offers features such as high-resolution imaging, 3D/4D capabilities, and automated measurement tools that streamline the diagnostic process.

AI Innovations: The website also provides insights into the latest AI innovations in ultrasound technology. These innovations include automated image analysis, which can help reduce the time required for image interpretation and improve diagnostic consistency. By leveraging AI, practitioners can enhance their workflow and focus more on patient care.

**Member Benefits:** Membership in the Voluson™ Club comes with a host of exclusive benefits that are designed to support continuous learning and professional development. Members gain access to a variety of tools and resources that can enhance their clinical practice.

**Application Tips:** The club offers practical application tips that help practitioners make the most of their Voluson systems. These tips cover a range of topics, from optimizing image quality to using advanced features effectively.

**Clinical Image Galleries:** Members can access extensive clinical image galleries that showcase a wide range of ultrasound findings. These galleries serve as a valuable reference for practitioners, helping them to recognize and diagnose various conditions.

**Online Training Sessions:** The Voluson™ Club hosts online training sessions and webinars led by leading experts in fetal medicine. These sessions provide opportunities for continuous learning and staying updated with the latest advancements in the field. Topics covered in these webinars include advanced imaging techniques, new diagnostic protocols, and case studies.

**Special Offers:** Members are also informed about new products and special offers, allowing them to take advantage of the latest technologies and innovations at competitive prices.

### Practical Applications

The resources available on the Voluson™ Club website have numerous practical applications that can significantly enhance the efficiency and effectiveness of clinical practice.

**Report Writing:** The website provides standardized reporting formats and templates that streamline the documentation process. For example, the "OB/GYN Report Templates" section offers customizable templates that can be used to create comprehensive and consistent reports. These templates ensure that all relevant information is included and presented in a clear and organized manner.

**Patient Education:** Educating patients about their ultrasound findings and procedures is an important aspect of fetal medicine. The Voluson™ Club offers a variety of educational materials and visual aids that can help practitioners explain complex medical information to patients. The "Patient Education" section includes brochures and videos that can be shared with patients to help them understand their diagnosis and treatment options.

**Clinical Guidelines:** Staying updated with the latest clinical guidelines and best practices is crucial for providing high-quality care. The Voluson™ Club provides access to guidelines from reputable organizations such as the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG). These guidelines cover a wide range of topics, from screening protocols to diagnostic criteria, ensuring that practitioners have the most current information at their fingertips.

### Why Visit This Website?

The Voluson™ Club is a valuable resource for fetal medicine practitioners for several reasons:

**Standard Reporting Formats:** The availability of standardized reporting formats and templates helps streamline the documentation process, ensuring consistency and accuracy in patient records.

**Up-to-Date Calculators:** The website offers up-to-date calculators for various measurements and assessments, aiding in accurate and efficient clinical decision-making.

**Prenatal Risk Assessment Tools:** Access to prenatal risk assessment tools allows practitioners to evaluate the risk of various conditions and make informed decisions about patient care.

**Comprehensive Educational Resources:** The extensive range of educational resources available on the website supports continuous learning and professional development.

Exclusive Member Benefits: Membership in the Voluson™ Club provides access to exclusive tools, resources, and special offers that enhance clinical practice and patient care.

In conclusion, the Voluson™ Club is an indispensable hub for fetal medicine practitioners. It offers a wealth of resources that support clinical practice, research, and education, making it a valuable tool for anyone in the field. Whether you are looking to stay updated with the latest advancements, improve your diagnostic skills, or enhance patient education, the Voluson™ Club has something to offer.

Some other similar websites from other leading ultrasound manufacturers that offer valuable resources for us:

1. **Siemens Healthineers - Siemens Healthineers Ultrasound Community**

   **Website:** ***https://www.siemens-healthineers.com***

   **Overview:** Siemens Healthineers provides a comprehensive online community for ultrasound practitioners. The platform offers a variety of educational resources, including webinars, case studies, and clinical guidelines. Members can access detailed information about Siemens' ultrasound products, such as the ACUSON Sequoia and ACUSON Juniper systems, which are known for their advanced imaging capabilities and user-friendly interfaces.

   **Key Features:**

   **Educational Webinars:** Regularly hosted by experts in the field

   **Case Studies:** Real-world examples to enhance diagnostic skills

   **Clinical Guidelines:** Access to the latest best practices and protocols

2. **Philips Healthcare - Philips Ultrasound Learning Center**

   **Website:**
   ***https://www.learningconnection.philips.com/en/ultrasound-education***

   **Overview:** Philips Healthcare offers an extensive learning center dedicated to ultrasound education. The platform includes a range of resources such as e-learning modules, clinical white papers, and product tutorials. Philips' ultrasound systems, like the EPIQ and Affiniti series, are highlighted for their innovative features and clinical applications.

**Key Features:**

**E-Learning Modules:** Interactive courses covering various ultrasound topics

**Clinical White Papers:** In-depth research articles and studies

**Product Tutorials:** Step-by-step guides to using Philips ultrasound systems

3. **Fujifilm Sonosite - Sonosite Institute**

**Website:** ***https://www.sonosite.com/in/education/sonosite-institute***

**Overview:** The Sonosite Institute by Fujifilm Sonosite is an educational platform designed for ultrasound practitioners. It offers a wealth of resources, including video tutorials, clinical case studies, and interactive learning modules. The institute focuses on point-of-care ultrasound (POCUS) and provides insights into the use of Sonosite's portable ultrasound systems.

**Key Features:**

**Video Tutorials:** Demonstrations of ultrasound techniques and procedures

**Clinical Case Studies:** Examples of ultrasound applications in various clinical settings

**Interactive Learning Modules:** Engaging content to enhance learning

4. **Canon Medical Systems - Canon Medical Ultrasound Academy**

**Website:** ***https://us.medical.canon/education/canon-medical-academy/***

**Overview:** Canon Medical Systems offers the Ultrasound Academy, a platform dedicated to ultrasound education and training. The academy provides access to webinars, clinical case studies, and product information. Canon's Aplio and Xario series are featured for their advanced imaging technologies and clinical versatility.

**Key Features:**

**Webinars:** Live and recorded sessions on various ultrasound topics

**Clinical Case Studies:** Detailed examples to improve diagnostic accuracy

**Product Information:** Insights into the latest ultrasound technologies from Canon

5. **Mindray - Mindray Ultrasound Education**

**Website:** **https://www.mindray.com/in/services/service-solutions/training-and-education/**

**Overview:** Mindray's ultrasound education platform offers a range of resources for practitioners, including online courses, clinical case libraries, and product training. Mindray's Resona and DC series are highlighted for their innovative features and clinical applications.

**Key Features:**

**Online Courses:** Comprehensive training on ultrasound techniques and applications

**Clinical Case Libraries:** Access to a wide range of case studies

**Product Training:** Detailed guides on using Mindray ultrasound systems

6. **Samsung Medison - Samsung Ultrasound Education**

**Website:** ***https://samsungmedison.com/***

Overview: Samsung Medison provides an educational platform for ultrasound practitioners, featuring webinars, clinical case studies, and product information. Samsung's WS80A and HS70A systems are known for their advanced imaging capabilities and user-friendly interfaces.

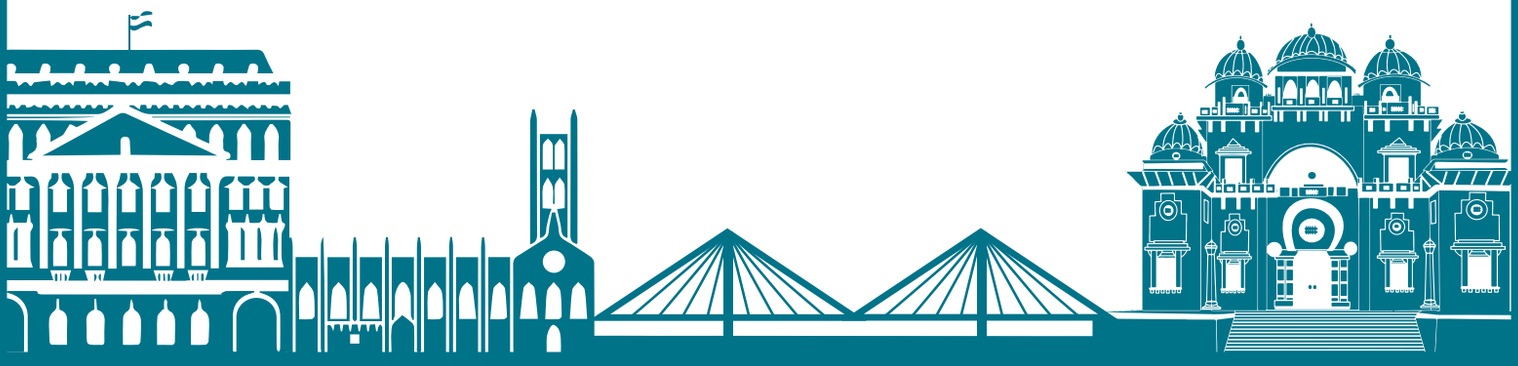
**Key Features:**

**Webinars:** Educational sessions led by industry experts

**Clinical Case Studies:** Real-world examples to enhance diagnostic skills

**Product Information:** Detailed insights into Samsung's ultrasound technologies

These websites offer a wealth of resources that can help ultrasound practitioners stay updated with the latest advancements, improve their diagnostic skills, and enhance patient care. Each platform provides unique features and content tailored to the needs of medical professionals in the field of ultrasound.



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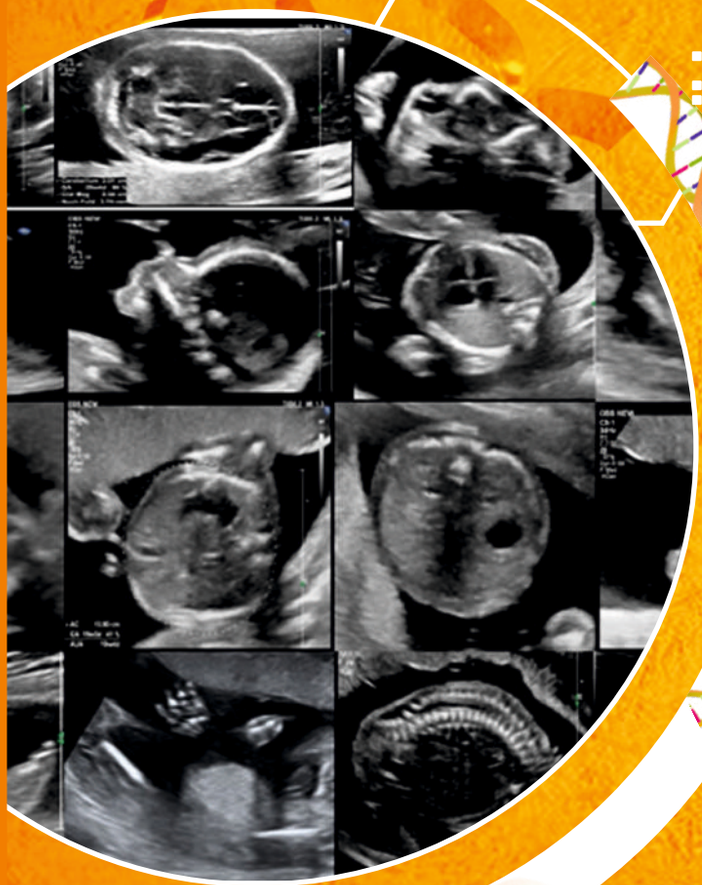

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# Online Mendelian Inheritance in Man (OMIM)

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## Introduction

OMIM is a comprehensive and authoritative database that catalogs human genes and genetic disorders. Established by Dr. Victor McKusick in 1966, OMIM serves as a crucial resource for geneticists, researchers, and clinicians, facilitating the understanding of the genetic basis of human diseases and the patterns of inheritance associated with various traits. Online Mendelian Inheritance in Man (OMIM®) is a continuously updated with particular focus on the molecular relationship between genetic variation and phenotypic expression. OMIM is based on the peer-reviewed biomedical literature, and criteria for inclusion of papers continue to evolve. In general, priority for inclusion is given to papers that provide significant insight into the gene-phenotype relationship, expand our understanding of human biology, or contribute to the characterization of a disorder. Information in each OMIM entry is cited, and the full reference is provided. OMIM is biocurated at the McKusick-Nathans Institute of Genetic Medicine, The Johns Hopkins University School of Medicine.

## Main features

OMIM is structured around a unique numerical system that assigns a distinct number to each entry, known as an OMIM number. These numbers typically take the form of a six-digit code, where the first digit indicates the major category of the disorder. For example:

| | |
|---|---|
| **1 ----- (100000-) 2 ----- (200000-)** | **Autosomal loci or phenotypes (entries created before May 15, 1994)** |
| **3 ----- (300000-)** | **X-linked loci or phenotypes** |
| **4 ----- (400000-)** | **Y-linked loci or phenotypes** |
| **5 ----- (500000-)** | **Mitochondrial loci or phenotypes** |
| **6 ----- (600000-)** | **Autosomal loci or phenotypes (entries created after May 15, 1994)** |

Allelic variants (mutations) are designated by the MIM number of the entry, followed by a decimal point and a unique 4-digit variant number. For example, allelic variants in the factor IX gene (300746) are numbered 300746.0001 through 300746.0101

This systematic numbering allows users to quickly identify the nature of a genetic condition and its inheritance pattern. The use of these numbers greatly enhances the utility of OMIM. The symbols used in OMIM serve various purposes to help users navigate and the information presented. Following are the examples:

An **asterisk (*)** before an entry number indicates a gene.

A **number symbol (#)** before an entry number indicates that it is a descriptive entry, usually of a phenotype, and does not represent a unique locus. The reason for the use of the number symbol is given in the first paragraph of the entry. Discussion of any gene(s) related to the phenotype resides in another entry(ies) as described in the first paragraph.

A **plus sign (+)** before an entry number indicates that the entry contains the description of a gene of known sequence and a phenotype.

A **percent sign (%)** before an entry number indicates that the entry describes a confirmed mendelian phenotype or phenotypic locus for which the underlying molecular basis is not known.

No **symbol** before an entry number generally indicates a description of a phenotype for which the mendelian basis, although suspected, has not been clearly established or that the separateness of this phenotype from that in another entry is unclear.

A **caret (^)** before an entry number means the entry no longer exists because it was removed from the database or moved to another entry as indicated.

When searching for information, users can input the OMIM number directly into the search bar, leading them to the corresponding entry. Each entry contains detailed information about the genetic condition, including its clinical features, inheritance patterns, associated genes, and references to relevant scientific literature. This structured approach makes it easier for users to navigate the vast amount of data and find specific information on genetic disorders. In addition to the main OMIM number, sub-numbers may be assigned to specific mutations or variants within a gene, providing even more granular information. For instance, a primary entry for a particular disorder may contain multiple sub-entries that detail different mutations associated with that condition. This allows for a more nuanced understanding of how genetic variations can lead to diverse phenotypes, even within the same disorder OMIM

also addresses the complexities of genetic disorders that do not follow classic Mendelian inheritance patterns.Some entries are designated as "phenotypic series," indicating that multiple genetic causes may contribute to a similar clinical presentation. This classification highlights the importance of considering both genetic and environmental factors in the expression of certain traits OMIM's impact extends beyond research and clinical practice. It plays a vital role in education as well.

### Practical application

The database is widely used in academic settings, where students and educators can access up-to-date information on genetics and hereditary conditions. By providing a centralized source of knowledge, OMIM supports the training of future geneticists and healthcare professionals, ensuring they are well-informed about the latest developments in the field Moreover, OMIM is continuously updated to reflect new discoveries and advancements in genetics. A dedicated team of curators reviews the literature, incorporating new findings into the database and ensuring that the information remains accurate and relevant. This commitment to quality control is critical, as the field of genetics is rapidly evolving, with new genes and variants being identified regularly.

In conclusion, Online Mendelian Inheritance in Man (OMIM) is an indispensable resource for anyone interested in the genetics of human diseases. Its systematic numerical classification system not only facilitates easy navigation of the database but also enhances the understanding of the inheritance patterns of various disorders. By providing comprehensive and up-to-date information on genes and their associated conditions, OMIM continues to play a pivotal role in advancing genetic research, clinical practice, and education in the field of human genetics. As the understanding of genetics deepens, OMIM will remain a cornerstone for researchers and clinicians alike.

## Dive Deeper

OMIM

Brief video overview of how to search OMIM
MIMmatch video tutorial

# Genereviews – A Key Resource for Genetics, Genetic Counseling and Fetal Medicine

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GeneReviews, hosted by NCBI (National Center for Biotechnology Information) maintained by a team led by Roberta A. Pagon, MD at the University of Washington, is a compendium of continually updated comprehensive online resource providing expert-authored and peer-reviewed disease descriptions that relate genetic testing to the diagnosis, management and genetic counseling of patients and families with specific inherited conditions. Additionally it provides clinically actionable information about inherited genetic conditions, serving as a key point-of-care tool for healthcare professionals. GeneReviews is the go-to site for quick answers and summaries pertaining to all of the major disorders subject to molecular genetic testing and also includes chromosomal abnormalities. For those in the field of fetal medicine, it provides valuable insights into how genetic conditions can impact prenatal care and fetal development, supporting informed decision-making in clinical practice.

**Key features of GeneReviews:**

### Comprehensive Coverage:

GeneReviews currently comprises 911 chapters and has more than ten million users annually. It covers a wide range of genetic diseases, with chapters focused on individual genes or phenotypes, as well as overviews summarizing causes of common genetic conditions like hearing loss or Alzheimer's disease.

### Expert Authorship:

Each GeneReview chapter is written by one or more specialists in the specific genetic condition, ensuring accuracy and in-depth knowledge. To ensure continuing relevant and medically actionable content, each GeneReviews chapter is updated every four to five years (or as needed) by the author(s) in a formal and comprehensive process curated by the GeneReviews editors.

### Standardized Format:

Presents information in a consistent journal-style format, making it easy to navigate and compare across different genetic disorders.

### Accessibility

GeneReviews articles freely are searchable by disease name. gene symbol, protein name, author, or tile. It is accessible through the NCBI Bookshelf platform, allowing broad access to medical professionals globally. GeneReviews are indexed in PubMed.

**Content Highlights:**

1. **Genetic Disorder Overviews:** Genereviews offers detailed, peer-reviewed articles on a wide range of genetic conditions, including those with a significant impact on fetal development. For fetal medicine practitioners, understanding these conditions is crucial for prenatal diagnosis and counseling. The platform covers both common and rare genetic disorders, providing a wealth of information on etiology, clinical presentation, and management strategies relevant to pregnancy.
2. **Inheritance Patterns and Genetic Mechanisms:** The website provides clear explanations of inheritance patterns such as autosomal dominant, autosomal recessive, X-linked, and mitochondrial inheritance. For fetal medicine specialists, understanding these patterns is vital when assessing the risk of genetic disorders in fetuses, particularly in the context of family history and prenatal screening results.

3. **Clinical Guidelines and Management:** Genereviews includes up-to-date, evidence-based clinical guidelines for managing genetic conditions. These guidelines are particularly beneficial for fetal medicine practitioners who often encounter genetic conditions in prenatal screenings and diagnostic imaging. The site provides recommendations for the management of these conditions, ensuring that practitioners adhere to best practices in prenatal care.
4. **Diagnostic Approaches and Testing:** The website outlines various genetic diagnostic tools and techniques that can be utilized during pregnancy, such as amniocentesis, CVS (chorionic villus sampling), and non-invasive prenatal testing (NIPT). These resources are indispensable for fetal medicine practitioners who need to select and interpret the appropriate diagnostic tests based on individual patient cases.
5. **Case Studies and Clinical Applications:** Genereviews includes real-world case studies that illustrate how genetic conditions are managed in clinical practice, offering insights into complex prenatal cases. For fetal medicine professionals, these case studies provide invaluable examples of how genetic counseling and testing are applied to prenatal care, helping practitioners develop informed management plans for pregnant patients.
6. **Educational Materials and Resources:** For professionals seeking to expand their knowledge, Genereviews offers educational materials including interactive modules, webinars, and articles. These resources are highly beneficial for fetal medicine practitioners who wish to stay current with the evolving field of genetics, prenatal diagnosis, and genetic counseling.
7. **Access to Published Research:** Genereviews offers links to a wide range of research articles and clinical studies, allowing fetal medicine practitioners to access the latest findings on genetic conditions and prenatal care. Staying updated on new genetic research is essential for improving clinical outcomes in fetal medicine, particularly when managing complex cases involving genetic anomalies.

## Practical Applications:

- **Clinical Practice:** For fetal medicine practitioners, Genereviews is a valuable tool for understanding genetic conditions that may impact prenatal care and fetal development. The website's comprehensive articles on genetic disorders, clinical guidelines, and diagnostic strategies help inform clinical decision-making and ensure that practitioners are following evidence-based practices when managing complex prenatal cases.
- **Patient Education:** Genereviews serves as a key resource for educating patients about genetic risks during pregnancy. The site's clear, well-organized explanations of genetic conditions and inheritance patterns make it easier for fetal medicine professionals to communicate complex genetic information to expectant parents, helping them make informed decisions about prenatal testing and care.
- **Report Writing:** Fetal medicine practitioners can use the wealth of information available on Genereviews to support their report writing. Whether it's documenting a diagnosis, preparing a prenatal risk assessment, or outlining a management plan, the detailed genetic and clinical data on the website provides a strong foundation for preparing thorough, accurate reports for patients, colleagues, or referring specialists.
- **Research and Continuing Education:** For those involved in research or ongoing professional development, Genereviews is an indispensable resource. Fetal medicine practitioners can access the latest research studies on genetic disorders and prenatal care, keeping them informed about advancements in genetics and how they may influence prenatal diagnosis and treatment strategies. Additionally, the educational tools and webinars provide opportunities for continuing education, enabling practitioners to stay up-to-date with the latest developments in the field.

## Why Visit This Website:

- Comprehensive, expert reviewed articles on genetic conditions with relevance to prenatal care
- Detailed explanations of inheritance patterns, crucial for assessing genetic risks in fetuses
- Up-to-date clinical guidelines for managing genetic disorders during pregnancy
- Information on the latest diagnostic tools and technologies for prenatal genetic testing
- Case studies and real-word applications of genetic counseling, and prenatal diagnosis
- Educational resources for continuing education and professional development
- Access to the latest research publications and clinical trials in genetics and fetal medicine

Genereviews is a vital resource for fetal medicine practitioners, providing detailed, up-to-date information on genetics and prenatal care. By offering expert-reviewed articles, clinical guidelines, diagnostic strategies, and educational materials, it supports the work of professionals in genetic counseling, prenatal diagnosis, and patient care. Whether for clinical decision-making, patient education, or staying current with research, Genereviews is an indispensable tool in the field of fetal medicine.Genereviews is a vital resource for fetal medicine practitioners, providing detailed, up-to-date information on genetics and prenatal care. By offering expert-reviewed articles, clinical guidelines, diagnostic strategies, and educational materials, it supports the work of professionals in genetic counseling, prenatal diagnosis, and patient care. Whether for clinical decision-making, patient education, or staying current with research, Genereviews is an indispensable tool in the field of fetal medicine.

## References:


1. Pagon, Roberta A.; Tarczy-Hornoch, Peter; Baskin, Patricia K.; Edwards, Joseph E.; Covington, Maxine L.; Espeseth, Miriam; Beahler, Christine; Bird, Thomas D.; Popovich, Bradley (2002). "GeneTests-GeneClinics: Genetic testing information for a growing audience". *Human Mutation*. **19** (5): 501–9. doi:10.1002/humu.10069. PMID 11968082. S2CID 23996040.
2. "GeneReviews". Retrieved 2013-06-13.


# Face2Gene: Revolutionizing Syndrome Recognition Through AI

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**Face2Gene** is an advanced AI-powered clinical tool designed to assist healthcare professionals in diagnosing genetic disorders through facial analysis. Developed by FDNA, this platform utilizes deep learning and pattern recognition to compare patient facial features with a vast database of known syndromes, helping geneticists and clinicians narrow down potential diagnoses. By integrating dysmorphology, genomics, and AI, Face2Gene enhances diagnostic accuracy, particularly for rare genetic conditions. While not a standalone diagnostic tool, it serves as a valuable aid in clinical assessments, improving early detection and personalized care for patients with genetic syndromes

Face2gene offers a user friendly interface, and the platform is quite easy to use

### Step 1: Accessing the Platform

Visit Face2Gene on a web browser.

Sign up or log in using your registered clinician credentials (only healthcare professionals can access the tool).

### Step 2: Uploading a Patient Image

Click on "New Case" to start an analysis.

Upload a clear, front-facing photo of the patient's face.

Ensure good lighting and no obstructions (e.g., glasses, hair covering features).

The AI will process the image and extract facial phenotypic features.

### Step 3: AI-Powered Syndrome Analysis

DeepGestalt™ AI compares the uploaded image to its extensive genetic syndrome database.

The system generates a ranked list of possible syndromes based on facial similarity.

Each suggested syndrome comes with a similarity score and links to relevant clinical information.

### Step 4: Adding Clinical & Phenotypic Data

Enter patient details (age, sex, ethnicity) for more accurate results.

Input additional phenotypic features using Human Phenotype Ontology (HPO) terms.

The system refines its suggestions based on this added clinical data.

### Step 5: Reviewing and Refining Diagnosis

Explore the top-ranked syndromes and their descriptions.

Click on a syndrome to view more details, including associated symptoms, genetics, and references.

Compare patient features with documented cases in the Face2Gene database.

### Step 6: Collaboration & Case Sharing

Save the case for future reference.

Share anonymized cases with colleagues or expert groups for a second opinion.

Use the platform's educational resources to compare cases and improve dysmorphology recognition skills.

### Step 7: Next Steps for Diagnosis

If a likely syndrome is identified: Consider confirming with genetic testing (e.g., whole-exome sequencing, microarray).

If uncertain: Use the differential diagnosis list to explore alternative possibilities or consult with genetic experts.

Below are the different types of forums available on the Face2Gene website:

### 1. Clinical Forums

- **Purpose:** These forums are designed for healthcare professionals to discuss clinical cases, share diagnostic challenges, and seek advice from peers.
- **Features:**
  - Case discussions with anonymized patient data.
  - Expert opinions and second opinions on complex cases.
  - Collaboration on differential diagnoses and management strategies.
- **Audience:** Clinicians, geneticists, and other medical professionals.

### 2. Research Forums

- **Purpose:** These forums focus on the latest research in genetics and genomics, providing a space for researchers to share findings, discuss methodologies, and collaborate on studies.
- **Features:**
  - Discussions on recent publications and breakthroughs.
  - Opportunities for collaborative research projects.
  - Sharing of datasets and research tools.
- **Audience:** Researchers, academicians, and scientists in the field of genetics.

### 3. Educational Forums

- **Purpose:** Aimed at fostering education and continuous learning, these forums provide resources and discussions on various topics in genetics and genomics.
- **Features:**
  - Webinars and online courses.
  - Discussion threads on educational topics and case studies.
  - Q&A sessions with experts.
- **Audience:** Medical students, residents, fellows, and educators.

### 4. Technical Support Forums

- **Purpose:** These forums offer technical assistance and support for users of the Face2Gene platform.
- **Features:**
  - Troubleshooting and technical issue resolution.
  - Updates on new features and platform enhancements.
  - User guides and tutorials.
- **Audience:** All users of the Face2Gene platform.

### 5. Community Forums

- **Purpose:** These forums are designed to build a sense of community among users, encouraging networking and informal discussions.
- **Features:**
  - General discussions on genetics and healthcare.
  - Networking opportunities with peers.
  - Announcements about conferences, events, and community initiatives.
- **Audience:** All members of the Face2Gene community.

### 6. Special Interest Groups (SIGs)

- **Purpose:** SIGs are specialized forums focused on specific areas of interest within genetics and genomics.
- **Features:**
  - Focused discussions on niche topics (e.g., rare diseases, cancer genetics).
  - Collaboration with experts in specific fields.
  - Resources and literature specific to the area of interest.
- **Audience:** Professionals with a specific interest in the topic of the SIG.

### 7. Patient Advocacy Forums

- **Purpose:** These forums provide a platform for patient advocacy groups and healthcare professionals to discuss patient care, advocacy efforts, and support strategies.
- **Features:**
  - Discussions on patient care and support.
  - Sharing of resources for patient advocacy.
  - Collaboration on initiatives to improve patient outcomes.
- **Audience:** Patient advocacy groups, healthcare providers, and caregivers.

### 8. Ethical and Legal Forums

- **Purpose:** These forums address the ethical and legal aspects of genetic testing and research.
- **Features:**
  - Discussions on ethical dilemmas in genetics.
  - Legal considerations in genetic testing and data sharing.
  - Policy discussions and updates.
- **Audience:** Ethicists, legal professionals, and healthcare providers.

1. **URL:** ***https://www.face2gene.com***
2. **5 Reasons to Visit Face2Gene:**

- Advanced AI-Powered Genetic Analysis: Face2Gene uses cutting-edge artificial intelligence to assist healthcare professionals in identifying rare genetic disorders based on facial features, aiding in faster and more accurate diagnoses.
- Comprehensive Clinical Decision Support: The platform provides tools and resources to help clinicians streamline their workflow, offering insights into potential genetic conditions and relevant medical literature.
- Global Collaboration: Face2Gene fosters a collaborative environment where clinicians and researchers worldwide can share knowledge and improve diagnostic outcomes for patients with rare diseases.
- Patient-Centric Approach: By enabling early and accurate diagnosis, Face2Gene helps improve patient care and quality of life, especially for individuals with rare genetic conditions.
- Free for Healthcare Professionals: The platform is available at no cost to qualified healthcare providers, making it an accessible tool for improving diagnostic accuracy and patient outcomes.

**Dive Deeper**

Face2Gene

# Phenotip: A Web-Based Tool for Prenatal Syndrome Diagnosis

**Dr Dipanjana Datta**
PhD, PDF
Genetic Counsellor, CGC,
Organization of Rare Disease India,
Renew Healthcare, Kolkata,
Institute of Fetal Medicine, Kolkata,
Institute of Child Health, Kolkata

## Introduction

Phenotip is a free, web-based tool developed to assist in the antenatal diagnosis of fetal syndromes through sonographic markers. It was designed to address the challenge of differentiating among the numerous syndromes with overlapping prenatal findings. The platform integrates a structured database of syndromic sonographic markers, providing a user-friendly interface for clinicians to identify potential diagnoses.

## Background and Need

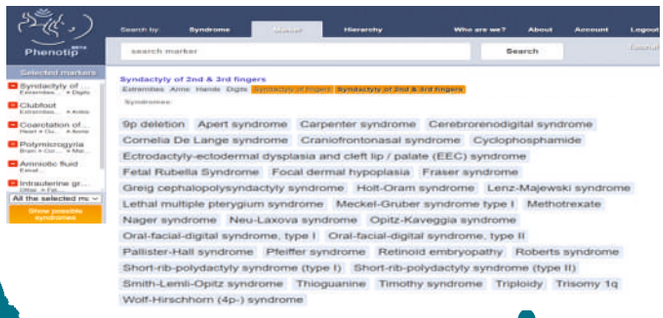
Prenatal ultrasound is a crucial tool in fetal anomaly detection. While some genetic syndromes can be confirmed through karyotyping or advanced genetic testing, many remain difficult to diagnose due to their complex phenotypic presentations. Existing genetic databases such as OMIM, Orphanet, and POSSUMweb, though valuable, do not specifically incorporate prenatal ultrasound findings. Phenotip was created to bridge this gap by offering a searchable database based solely on sonographic markers, aiding fetal medicine specialists, sonographers, and geneticists in their diagnostic process.

## Database Structure and Functionality

Phenotip utilizes a hierarchical tree of 1,140 sonographic markers, categorized by organ systems. Each marker is further divided into subcategories, enhancing search specificity. For example, the category "face" can be further broken down into "eyes," "ears," and "mouth," with subcategories such as "cleft lip," "micrognathia," or "low-set ears." Users can input one or multiple markers, and the system generates a list of syndromes associated with these findings. The tool also accommodates synonym recognition to ensure comprehensive search results.

## Searching the Database

The user can search for syndromes by entering specific ultrasound findings, which can be selected from an expandable tree or a direct search box. The system allows users to refine searches by adjusting specificity and sensitivity. Selecting broader markers increases sensitivity by considering all related sub-markers, whereas selecting more specific markers enhances diagnostic precision. Additionally, Phenotip suggests additional markers that may help differentiate between similar syndromes. For instance, if a user inputs "polydactyly" and "congenital heart defect," the tool might suggest evaluating for Ellis-van Creveld syndrome, which is known for these features.




## Validation and Results

Since its launch in 2013, Phenotip has been accessed by users across 18 countries, demonstrating its global applicability. A retrospective analysis of 50 cases from TheFetus.net showed that the tool provided the correct diagnosis in 86% of cases, with the remaining cases yielding a set of potential diagnoses that included the correct one. For example, in a case where ultrasound findings included "hydrops fetalis," "micrognathia," and "short long bones," Phenotip suggested possible syndromes such as Noonan syndrome and Lethal Multiple Pterygium syndrome, aiding in further genetic evaluation and testing.

## Future Enhancements

Phenotip is an evolving tool, with ongoing updates to expand the database and refine search algorithms. Future developments include the integration of MRI findings, prioritization of search results based on syndrome prevalence, and incorporation of postnatal findings to enhance diagnostic accuracy. Additionally, the system is expected to adapt Bayesian models to improve the probability-based ranking of diagnoses. For example, a syndrome with a higher prevalence rate and a closer match to multiple markers entered by the user would be ranked higher in the search results.

## Conclusion

Phenotip is a pioneering, freely accessible web tool that enhances the accuracy of prenatal syndrome diagnosis through structured sonographic marker analysis. While it does not replace expert evaluation, it serves as a valuable adjunct for clinicians navigating the complexities of fetal syndromic identification. With continuous updates and refinements, Phenotip has the potential to become an indispensable resource in fetal medicine, allowing clinicians to make more informed decisions regarding prenatal diagnosis and management.

**Dive Deeper**

PHENOTIP

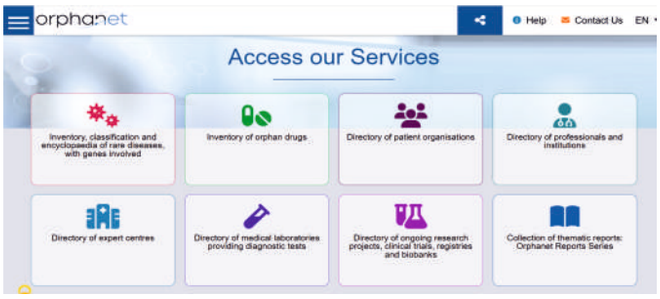
# Orphanet: A Key Resource for Rare Diseases

**Ms Sutopa Singha**
M.Sc, BGCI level 2 Certified genetic counsellor,
Medical Science Liaison & Genetic Counselor
Lilac Insights

## Introduction

Orphanet, established in France by INSERM (French National Institute for Health and Medical Research) in 1997 and later expanding into a global consortium of 40 countries, is a unique and vital resource dedicated to improving the lives of individuals affected by rare diseases. It achieves this by gathering, validating, and disseminating high-quality knowledge on these conditions, thereby enhancing diagnosis, care, and treatment. Orphanet's core mission is to provide equitable access to this information for all stakeholders worldwide, including patients, families, healthcare professionals, researchers, and policymakers. A key component of its work is the maintenance of the Orphanet rare disease nomenclature (ORPHAcode), a crucial tool for standardizing rare disease classification and improving their visibility within health and research information systems.

## Content Highlights




## Access to Services:

This section provides crucial resources for healthcare professionals, researchers, and patients, including:

- **Inventory, Classification & Encyclopedia of Rare Diseases:** A comprehensive repository of rare diseases, including their classification and associated genes.
- **Inventory of Orphan Drugs:** Information on drugs designated for the treatment of rare diseases, including their development status and availability.
- **Directory of Patient Organizations:** A global list of organizations supporting patients with rare diseases, offering advocacy, resources, and support networks.
- **Directory of Professionals and Institutions:** A catalog of institutions specializing in research, diagnostics, and treatment of rare diseases.
- **Directory of Expert Centres:** Listings of medical centers with specialized expertise in managing and researching rare conditions.
- **Directory of Medical & Laboratory Providers for Diagnostic Tests:** A registry of laboratories offering diagnostic tests for various rare diseases.
- **Directory of Ongoing Research Projects, Clinical Trials, Registries & Biobanks:** Access to research studies, clinical trials, and biological sample repositories focused on rare diseases.
- **Collection of Thematic Reports (Orphan Report Series):** A collection of reports providing insights into trends, advancements, and challenges in rare disease research and management.

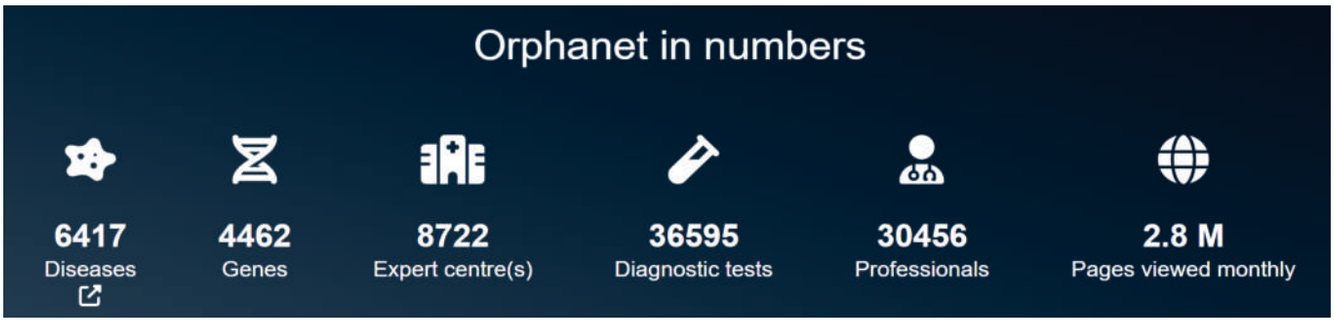
## Practical Applications in Fetal Medicine

- **Prenatal Diagnosis Support:** Orpha.net provides essential genetic and phenotypic information for prenatal screening and diagnosis of rare conditions.
- **Guideline-Based Management:** The website offers clinical guidelines for managing pregnancies affected by rare fetal anomalies.
- **Consultation and Referral:** The expert directory helps fetal medicine practitioners connect with specialists for interdisciplinary care.
- **Research and Case Reporting:** Facilitates access to research networks and rare disease registries, enabling clinicians to contribute to case studies and advancements in the field.
- **Patient Counseling:** Provides reliable information to support genetic counseling for expectant parents dealing with a rare disease diagnosis.

**Website URL: www.orpha.net**

**Why Visit This Website:**

- Comprehensive database on rare diseases relevant to fetal medicine, researchers, Clinical geneticists etc.
- Diagnostic and classification tools for complex fetal conditions.
- Up-to-date clinical guidelines and expert recommendations.
- Access to orphan drug databases and clinical trials.
- International directory of experts and specialized care centers.




This guide aims to highlight the utility of Orpha.net in fetal medicine, ensuring that practitioners have quick access to credible and updated information for patient care, research, and education.

**Dive Deeper**

Orphanet

# ClinVar: A Free Repository for Genomic Variations

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ClinVar is a freely available public archive/repository of genomic variations and corresponding phenotypes submitted by clinicians, clinical laboratories, researchers, expert panels or different organizations with available supporting evidences to interpret the clinical significance of the variant. This database is an integral part of Clinical Genome Resource (ClinGen) project, funded by NIH to understand relevance of genes and variants with respect to the disease, deciphering the pathogenicity of those variants. The database is maintained by National Center for Biotechnology information (NCBI). ClinVar is currently (Jan 20,2025) a powerful database of 4960010 submitted records, with 17561 genes' specific variants submitted by 2985 submitters. The variants can be of any type and length which include cytogenetic rearrangements, SNVs, small insertion/deletions, to copy number variation identified from germline or somatic origin, classified in Mendelian disorder, cancer or drug response category.

The submitter may include variant details based on a case with phenotypes along with additional evidences and literature citations supporting the interpretation or they can simply submit variant level minimal information, where ClinVar maps the variant to reference sequence, assign identifiers in the form of SCV format, and add other publicly available data aggregated for the variant. Variant accuracy evaluation or modification are not done by ClinVar after submission of a variant, however ClinVar assigns a review status to submissions which calculate the aggregate interpretation based on submitter type, expert review, consensus in the interpretation, transparency into the rules used for interpretation. The submitted variants can be classified as pathogenic, pathogenic/likely pathogenic, and conflicting interpretation of pathogenicity.

### Utility of Clinvar in practice

The ClinVar database can be searched by a specific HGVS expression, rs-ID, protein change, HGNC gene symbol, disease or phenotype. The search results can be filtered by classification type, germline classification, molecular consequences, types of conflicts, variation type, variation size, variant length and review status. Selecting a specific variant gives submitted data with available data aggregated by ClinVar. Thus, it helps users to explore different variants to understand their clinical significance. Clinvar provides an important evidence regarding already reported variants in a gene in affected cases which can help in variant reclassification based on the ACMG guidelines.

### Limitations

There are two major grey areas in ClinVar which leads to variant classification discordance, e.g conflicting interpretation of pathogenicity and Single submitter classification. Precautions should be taken while considering variants with conflicting interpretations of pathogenicity where multiple submitters provide different classification for the same variant. Since almost two third of the total variants in ClinVar are submitted by single submitter, the classification scheme by a single submitter can be unreliable, so further investigation for variant assessment will be beneficial. Additionally, as the novel variants are not immediately submitted to ClinVar, a huge number of variants still remains unknown to the users.

### Dive Deeper

CLINVAR NIH

### Clinvar

Every variant present in Clinvar may not be a disease causing variant and variants submitted by single or few submitters should be viewed with caution while using this evidence for variant reclassification

# DECIPHER - DatabasE of genomiC varlation and Phenotype in Humans using Ensembl Resources

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Hyderabad - 500033

**Synopsis:**

Genetic variations come in all sizes, these variations also have highly varying disease-causing potential. Because of this, the relationships between the variants & patient phenotypes may not be immediately apparent. Decipher is a user-friendly database that allows characterization of a genetic variant, so that it can be objectively & meaningfully interpreted in the context of patient features. It has two essential attributes – Clinical cases evaluated & curated by Clinical geneticists across the world and secondly, a wide array of tools which will compare & analyse the input variant. Designated clinical geneticists across various centres of the world are in-charge of the data quality & its curation.

## DECIPHER

DECIPHER was set up in 2004 by Nigel Carter of the Wellcome Trust Sanger Institute and Helen Firth, a clinical genetics consultant at Addenbrooke's Hospital in Cambridge. The website is currently run by the European Molecular Biology Laboratory, European Bioinformatics Institute (EMBL-EBI) and sponsored by Cambridge University Hospitals, NHS Foundation Trust. This interactive genomic tool has been receiving data since its inception in 2004 and comprises of variants interpreted several times over the last 20 years. The technologies identifying these variants have also evolved significantly.

The database has restricted public access – anonymized information relating to the variant & a summary of the phenotypes may be readily viewed. However, complete genotypic/ phenotypic data may not be accessible without the authorised consent of the centre, which maintains that data. The research or academic centres seeking to collaborate for such information may access the data with prior consent. Currently, Decipher has 50,019 open access patient records & it just keeps growing.

## Primary objectives

i. Distinguish between the benign & pathogenic variants based on various parametres

ii. Define the exact number of genes involved in a specific variant & predict their collective clinical consequence.

iii. Characterize novel gene/ disease or novel variant- disease relationships to aid in research which impacts diagnosis & treatment of genetic conditions. DECIPHER has been cited more than 2600 times in peer-reviewed publications.

In order to achieve the above, DECIPHER uses a wide array of external browsers (gnomAD, ClinVar & also sources information from other independently maintained datasets, which are thoroughly updated. Despite this seamless integration of multiple technologies & databases, absolute conclusions cannot be made in every case since it is subject to many biases. Some of them include testing resolution, clinical phenotyping bias especially concerning dysmorphic features, evolving nature of symptoms, penetrance, variable expressivity etc – to name a few!

## Key features

- The database is freely available online at https://www.deciphergenomics.org
- Genetic information is presented in the GRCh38 assembly version of the human genome to facilitate most up-to-date variant interpretation.
- GRCh37/hg19 co-ordinates can be mapped onto the GRCh38 assembly using a LiftOver tool.
- It supports almost all genomic variations – germline/ mosaic, nuclear/ mitochondrial genome, single nucleotide variations, copy number variations, repeat expansions & large structural variants.
- Large structural variants may be interpreted with caution as the breakpoints are only approximate
- Variations are classified as per international standards for variant classification
- Patient phenotypes are captured in Human Phenotype Ontology (HPO) terms, which allows computational comparison between patients.
- Variant pathogenicity claims are made by the submitter, based on the evidence available to them, in the context of their patients.
- The variant interpretation is dynamic since the website is incorporating latest information & is constantly evolving.
- Protein browser is available for protein-coding genes & maps the topographic effects of a gene variant on its corresponding protein.
- Variant pathogenicity claims are made by the submitter, based on the evidence available to them, in the context of their patients.

## Navigating the user-interface

- Let's understand how DECIPHER works through an example. A Chromosomal microarray report revealed the following variant - arr [GRCh38] Xq28(154264743_154450007) x 3

- Let's dismantle the components & understand what they mean!

  Arr  Type of technique used, microarray

  GRCh38  Version of Human reference genome used

  Xq28  'q' or the long arm of the X chromosome, region 2, band 8

  (154264743_154450007)  co-ordinates to indicate the portion of the chromosome involved

  x 3  Normal chromosomal complement is diploid (x2), 3 indicates duplication on one of the chromosome constituting a total of 3 copies of that segment in the patient.

- **Interpretation:** There is a heterozygous duplication of a segment spanning between the co-ordinates (154264743_154450007) on long arm of the chromosome X in the region 2, band 8.
- Log in to the DECIPHER database homepage. The general search option allows one to query by phenotype, gene, variant, disease, protein, pathogenicity, inheritance, consequence, etc.
- This page will lead to a list of previously deposited patient variants overlapping this region, the associated patient phenotypes, if there are any known CNV syndromes & the genes encompassed by this region.
- If the exact co-ordinates are known, like in this case, DECIPHER's Genome browser may be tried. The query has to be submitted in a specified way – X:154264743-154450007, for the variant mentioned above.
- Make sure that the correct reference version (GRCh37/ GRCh38) is used. If the variant is reported in GRCh37 version, then the query submitted should be grch37:X:154264743-154450007.
- Correct entry leads to a page where the variant is visually displayed with many trackers, aligned in a vertical manner.
- Each tracker gives specific information about the variant
  - i. Number of genes contained
  - ii. If any of those genes are morbid
  - iii. If a variant with exact same breakpoints have been reported & their phenotypes
  - iv. If not, whether a variant(s) closely matching in size are checked for.
    - It may be one single variant spanning the length of our variant.
    - Sometimes, it may include several smaller non-overlapping variants aligned horizontally making up the total size, in which case, the collective phenotype is obtained by summing up the individual features.
- The tracker comes with side buttons, which allow us to configure, visualize, and filter variants as required.
- One can systematically go through each of these trackers to learn several attributes of the variant, in order to gather evidence for pathogenicity/ benignity.
- Based on the information obtained, accurate counselling may be offered on the likely significance of the variants & its potential implications in the family.
- The interface also provides hyperlinks to the reported patient features, variant classification & relevant publications.
- There is a quick link to contact the corresponding clinician to access more details about the patient & initiate research.

## DECIPHER through pictures!

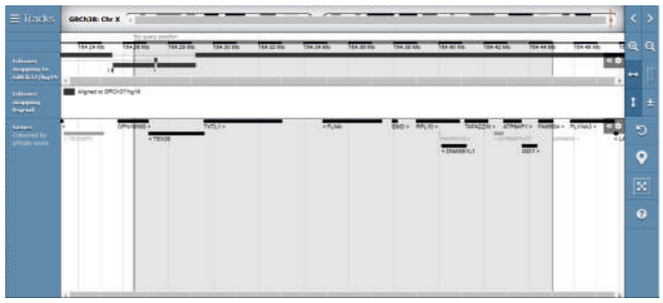

***Figure 1: Genome browser view for the X:154264743-154450007 variant. The top-most tracker shows the position on chromosome X in GRCh38 version, next tracker shows the same variant in GRCh37 view. The bottom tracker shows the number of genes contained in the duplicated segment. The density of the genes in this segment is poor. The individual genes & their significance may then be checked to understand the overall contribution towards the phenotype. The side buttons on the extreme right provide different viewing & filtering options.***

***Figure 2: The picture shows all the previously deposited duplicated variants (blue lines) in the DECIPHER overlapping this region. As is shown, there are no other duplication variants of the same size. Most of these are much larger variants, but they do overlap with our variant, either proximally, or distally, or both. Also note, our variant overlaps with a well-known Xq28 micro-duplication syndrome distally.***

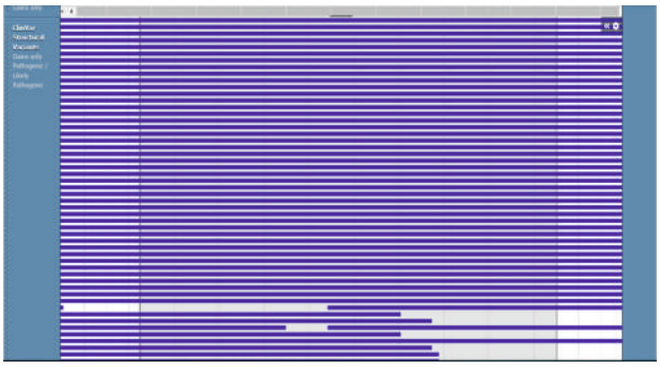

***Figure 3: Picture showing the reported larger duplication variants in ClinVar, almost completely encompassing our variant. The reported phenotypes may be due to the larger sizes of duplications, involving several other genes, which lie outside our variant. The contribution of the few genes in our variant cannot be deduced. Therefore, it is right to conclude that the exact significance of our variant remains unknown.***

## Current version

The latest version is version 11.29 of DECIPHER, which was released on 8 January 2025. This version boasts of new value additions such as –

a. **Cancer frequency data**
   Compiled by the National Disease Registration Service (NDRS) from diagnostic laboratories in England. Data comprises of 4,500 variants in 13 cancer susceptibility genes including BRCA1, BRCA2, MSH2, PTEN and SMAD4.

b. **Penetrance data for variants associated with cardiomyopathies** - VariantFX
   Population estimates of penetrance are shown alongside the allele frequencies, which are helpful especially when these variants are reported as secondary findings.

c. **Data on case/ control cohorts**
   Aids in assessing the strength of genotype-phenotype relationships in cardiac diseases. Information on canonical splice site variants and premature termination codon variants that undergo NMD are also available.

d. **Predictive scores for protein**
   Scores predict the probability of the possible disease mechanism associated with proteins such as dominant-negative, gain-of-function or loss-of-function mechanism. The literature evidence & the link to the supporting is also provided.

e. **gnomAD Short Tandem Repeat (STR) track**
   Provides data on 60 disease associated repeat loci with normal and pathogenic repeat lengths.

f. **Patient matches**
   Users can now search patients with similar genotypic-phenotypic profiles.

## Conclusion:

DECIPHER is a very user-friendly database, which now supports a wide range of human genetic variations & allows you to understand their significance in the context of patient phenotype. Plug the variants & play using. We welcome you to try out few variants from the Chromosomal Microarray report of your patients! Remember, the more you try, the interesting it gets…

## Dive Deeper

DECIPHER Mapping the Clinical Genome

# gnomAD: Aggregating Genetic Variations in the Population

**Ms Yashodhara Bhattacharya**
M.sc, CGC
Product Manager and
Sr. Genetic Counselor,
OneCell Diagnostics Pvt. Ltd.

## Background

DNA is the hereditary material present in all human beings. DNA comprises smaller segments known as the 'genes' which contain the code to produce different proteins in the body. The code of > 20,000 genes together produce all the proteins required to make a structural and functional human being.

Changes in the code of these genes do take place as part of evolution and are known as variations. Some variations or variants of the code may not lead to any significant change in the protein structure and thus may be benign, wherein others may lead to significant changes in the protein structure leading to medical symptoms. These variants are known as pathogenic or disease-causing variants.

Reference population databases are important in understanding the biological function of such genetic variation. The population frequency data helps distinguish rare variants which are likely to be causative of Mendelian disorders from benign variants which are commonly found across the human genome.

## Introduction

The Genome Aggregation Database or GnomAD is a population database for aggregating and harmonizing data from large scale sequencing projects (such as exome or genome sequencing worldwide) as reference for the scientific community. The database was earlier known as the Exome Aggregation Consortium (ExAC) dataset which was the first large scale aggregation of existing exome sequencing dataset from 60,000 individuals. With the addition of genome data it was renamed as Genome Aggregation Database and comprises variant data from over 1,95, 000 individuals. Variant data from such population databases are crucial for variant classification of clinical samples assisting in the diagnosis of Rare Diseases.

## Content Highlights

The GnomAD database in its current version v4.1.0 allows the user to search for a variant based upon the respective gene, region or variant details. In clinical practice, the genomic reports share the details regarding the variants identified in a patient. While reporting, the details of the gene variants shared including the gene, the locus of the variant (exon, coding sequence change), transcript ID. These details of the variant can be used to search for the population frequency of the variant in the GnomAD database.

The website gives the user the nomenclature to be used for defining the search based upon the requirement.


***Figure 1: Image of nomenclature for different variant searches in GnomAD***

### *Steps for using the GnomAD database:*

1. Go to the GnomAD website

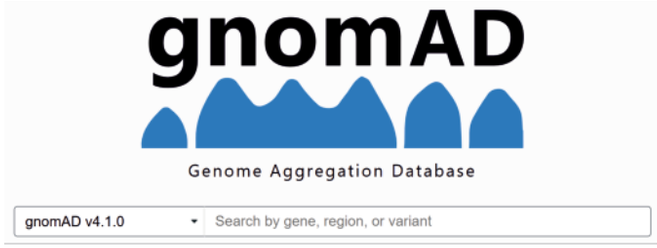


***Figure 2: Search bar in the gnomAD website***

2. In the search bar, the variant coordinates should be entered in the format given in Fig 1: [chromosome number]-[position]-[reference allele]-[alternate allele]

   Eg: 1-55051215-G-GA

3. The webpage displays information such as allele frequency in different populations, predicted functional consequence of the variant, and potentially related clinical data.

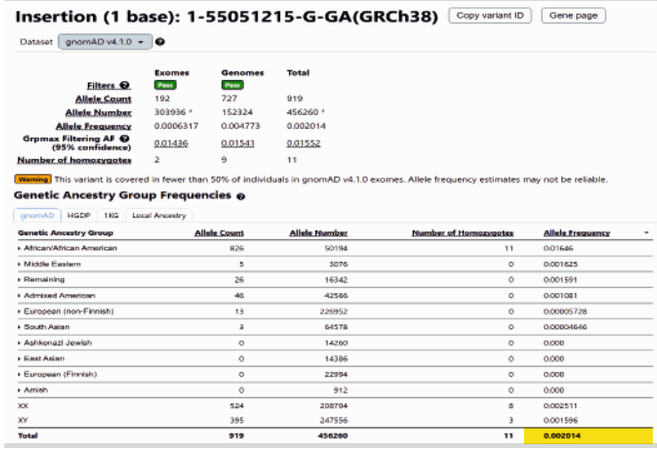


***Figure 3: Webpage displaying details of allele frequency in different populations***

**Highlighted value shows the allele frequency of the variant*

### Practical Application of using the GnomAD database:

Majority of pathogenic variants are rare and identifying such variants is a crucial step for analysis of Mendelian Rare Diseases. Based upon the ACMG guidelines the population allele frequency is an important criterion assisting in the classification of a variant.

- Prevalence of an allele in affected individuals compared to control (healthy) individuals is important evidence in support of pathogenicity
- A novel variant or an allele absent from the database may be considered moderate evidence of pathogenicity
- A variant with a high allele frequency or showing a strong presence in healthy individuals inconsistent with the disease penetrance may be considered evidence of being benign

For Fetal Medicine Consultants, the gnomAD database may be helpful in the interpretation of genetic test reports. An allele frequency of >1% is usually considered as benign and those with an allele frequency of <0.1% suggest a higher likelihood of being pathogenic. The data from the gnomAD database may give key information regarding the presence of an allele in a particular population assisting clinical correlation. However, it is important to remember that the allele frequencies may vary within different populations and certain variants may be pathogenic and show a high allelic frequency due to prevalence of the disease in the specific population. The allele frequency serves as important evidence in the classification of a variant based upon its likelihood of being pathogenic or being benign. However, it is a supporting criterion and often requires additional evidence while classifying a variant based upon ACMG guidelines.

### References:


- Chen, S.*, Francioli, L. C.*, Goodrich et al., A genomic mutational constraint map using variation in 76,156 human genomes. Nature. 625, 92-100 (2024). https://doi.org/10.1038/s41586-023-06045-0 PMID: 38057664
- Gudmundsson S, Singer-Berk M, Watts NA, Phu W, Goodrich JK, Solomonson M; Genome Aggregation Database Consortium; Rehm HL, MacArthur DG, O'Donnell-Luria A. Variant interpretation using population databases: Lessons from gnomAD. Hum Mutat. 2022 Aug;43(8):1012-1030. doi: 10.1002/humu.24309. Epub 2021 Dec 16. PMID: 34859531


### Why visit the website?

- GnomAD database is a publicly accessible collection of human genetic variation data
- It allows clinicians consulting individuals with rare genetic diseases compare and interpret genetic variants across diverse population
- This is important for identifying disease causing mutations in affected individuals and further estimating the risk of recurrence in subsequent generations
- It is also important for clinicians who may be involved in research studies with respect to genetic variation among different populations.
- Understanding the population based allelic frequency prevalence can also help create surveillance protocols for a specific population at high risk for having certain rare genetic diseases

### Dive Deeper


Genome Aggregation Database (GnomAD)

# Bengal SFM Activities:
# Where Knowledge Blooms and Collaboration Takes Flight!

The SFM Bengal recently organized a series of impactful events aimed at enhancing knowledge and collaboration in the medical community. One notable event was a Round Table Meeting (RTM) held in Bolpur, Shantiniketan, which welcomed esteemed dignitaries and fostered discussions on key healthcare issues.





SFM partnered with the Bengal Obstetrics and Gynaecological Society to host a scintillating webinar on January 28th, focusing on the prevention of birth defects. Dr. Prasanna Roy provided valuable insights into antenatal strategies to mitigate these risks, while Clinical Geneticist, Dr. Kausik Mandal addressed the genetic aspects and clinical diagnosis of birth defects.

The session culminated in a comprehensive panel discussion on correctable anomalies, moderated by Dr Seetha Ramamurthy Pal and Dr. Kanchan Mukherjee, featuring a multidisciplinary team of experts, including Dr. Ananya Basu, Dr. Bratati Bhattacharyya, Dr. Nandini Chakraborty, Dr. Priyanka Pipara, Dr. Shankar Dey, Dr. Sumona Haque, Dr. Mahua Roy, along with paediatric cardiologists and genetic counsellor Dr. Dipanjana Datta.


Additionally, On January 31st, SFM led an engaging webinar that spanned 5 to 10 weeks, featuring a distinguished panel that included Dr. Ashok Khurana, Dr. Kusargadhi Ghosh, Dr Ananya Basu, Dr. Arkajyoti Mukherjee, Dr. Seetha Ramamurthy Pal, and Dr. Prasanna Roy. This initiative was met with great enthusiasm from participants, highlighting the commitment to ongoing education in the field.

**Friday | 31st January, 2025 | 8:15 pm onwards**

| Time | Topic | Speaker |
|---|---|---|
| 08:15 pm - 08:20 pm | **Admittance of Attendees and Trade Partner Videos** | |
| 08:20 pm - 08:25 pm | Welcome Address | Dr. Seetha Ramamurthy Pal |
| 08:25 pm - 09:25 pm | **Panel Discussion:** 5-10 Weeks Scan : Scope and Limitations **Panelists:** Dr. Ashok Khurana, Dr. Kusagradhi Ghosh, Dr. Seetha Ramamurthy Pal, Dr. Prasanna Roy, Dr. Arkajyoti Mukherjee, Dr. Ananya Basu | **Moderator:** Dr. K Aparna Sharma |
| 09:25 pm - 09:30 pm | Trade Partner Videos | |
| 09:30 pm - 09:40 pm | Audience Interaction | Dr. Prasanna Roy |
| 09:40 pm - 09:45 pm | Vote of Thanks | Dr. Prasanna Roy |

These events exemplified SFM Bengal's dedication to advancing medical knowledge and interdisciplinary collaboration in maternal and child health.

8TH JUNE

# SAVE THE DATE

2025

# SFM BENGAL CHAPTER

## ANNUAL CONFERENCE 2025

Date: 8th June 2025 | Venue: Kolkata

**Further UPDATES to Follow**

# And finally...

## From the FM Radio

### Dear Fetal Fanatics,

Welcome to the very first edition of this column where our shared love for the tiniest, tiddliest humans would be celebrated with humour, humility and an occasional dose of delirium to keep us sane.

The Bengal Chapter proudly presents yet another spectacular issue of their theme-based newsletter. This time, we turn to World Wide Web—the elixir of prudent practice. In fetal lexicon we call it the Womb Wise Web. Will it transform you into a fetal medicine Savant overnight? Nope. Does it have that magic mantra for Hogwarts Wizardry? Obviously not. But it may elevate some standards!

Let's now take a little nostalgic look into our madness. Let's ask ourselves that primal but profound question of why we chose fetal medicine. Why not specialties where one could rise to fame in no time? Why not those areas where money would erupt like a volcano? The answer dear friends, lies in our peculiar, sometimes pathological tryst with the uncharted world of the womb. We are those funny old geeks who willingly trade happy hours for endless scans or for decoding rarest of the rare genetic syndromes. We love deciphering fetal anatomy like archaeologists unearthing valuable artefacts. Where others see blobs, we see brilliance. While others read Hebrew, we uncover the code of life. Fetal medicine is not just a specialty, it's a calling. Here precision marries intuition, science meets compassion and care goes beyond diagnosis. We are the Sherlocks of the nest, piecing together cryptic clues from shadowy pixels and unfathomable biochemistry reports.

But the pursuit is not that dreamy all the time. It has its own sweet challenges too. Remember the first few days of staring at the screen. Didn't it feel like the good old days of watching the b&w televisions in 80s with a crow conveniently perching on the rooftop rickety antenna messing up the signals? Or the initial attempts of NT scans? The probe slipped, fetus flipped and your shoulder flopped. You sat there downcast, with a lingering spirit whispering through the walls, Ni-co-lai-des! Yet, here we are, seasoned professionals, armed with smarter machines, *majboot hands lekin no majboori* in life!

Many of us took a holy fetal dip in our local lily pools, may be in a dingy diagnostic centre years back. We could well have been doing okay there but only as scattered dots. But then, like a beacon of hope came an IIT Baba (I mean the special one who traded his *pukka* IIT seat for MBBS at AIIMS) and joined the dots. The Maha Sangam of fetal minds came into being, in the name of SFM. The lone rangers got transformed into fetal avengers. Our own Kumbh Melas started happening. Well, isn't it time to raise a metaphorical toast to our mentor emeritus? Take a bow Dr Khurana. Without your visionary leadership, many of us would still be wandering the hospital corridors, ultrasound probe in hand, muttering away, "what the hell....!"

Finally a crisp invite for our next Kumbh in FetalBasicon Avatar, where you would meet the real Gurus at Gurugram and take a Shah-i-Snan with our beloved Dr Shah, the King of Kings.

That's all for today folks. Keep scanning. Keep laughing. Keep being extraordinary. Tune in again in a quarter at fetal frequency.

Truly yours
**The Fetus Uncle**

*Where Science Meets Satire*