

Foetus as a patient: Art and science of foetal medicine

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Foetal medicine encompasses the assessment of foetal health, and the maintenance of foetal growth and wellbeing. It requires the expertise of a foetal medicine specialist and input from various specialists, including among others obstetricians, maternal medicine specialists, neonatologists, paediatric cardiologists, paediatric surgical specialists, geneticists and specialist nursing counselors. The foetus therefore is considered by the professionals and parents as a potential patient in her or his own right. Foetal medicine is the specialty that addresses this unborn patient.

The progress made in foetal medicine has been notable over the period of time. Innovation in prenatal screening in early first-trimester for aneuploidy fetuses and for prediction of preeclampsia has improved the detection rate to 95% and 80% with a false-positive rate of 2% and 10%, respectively.¹ Recent developments in fluorescence in-situ hybridization or quantitative fluorescence PCR have led to rapid results within two-six hours of amniocentesis for trisomy 21.² Pre-implantation genetic diagnosis is now a well established technique for early prenatal diagnosis of chromosomal abnormalities in in-vitro fertilization pregnancies and as an alternative method for prenatal diagnosis in those families known to be at increased risk of having a child with certain severe single gene disorders.³

Advance in imaging with introduction of foetal magnetic resonance imaging is considered useful in particular for assessment of central nervous system abnormalities.⁴ Three- and four dimensional ultrasonography provides added information for the analysis of skeletal anomalies, facial malformations and assessment of neural tube defects.⁵ The technology has now offered new opportunities to study embryonic and foetal behaviour which would make information available on specific movement patterns and quality of movement in the high-risk foetus for detection of foetal neuro-developmental impairment.⁶

Until recently, the only question raised by the prenatal diagnosis of a foetal malformation was whether to abort the foetus or to await delivery. Now, treatment of several foetal disorders has proved to be feasible in-utero. With the recent advancement in minimally invasive foetal surgery, frequency of open hysterotomy has decreased considerably. The latter is performed for conditions such as myelomeningocele, large

congenital cystic adenomatoid malformation of lungs or a large sacrococcygeal teratoma.⁷ Prenatal repair of foetal myelomeningocele is now currently under investigation through a more systematic and appropriate randomized trial sponsored by National Institute of Child Health and Human Development.⁸ Introduction of minimally invasive technique for laser photocoagulation for the treatment of twin-twin transfusion syndrome,⁹ ablation of posterior urethral valves causing bladder outlet obstruction¹⁰ and tracheal occlusion using a balloon catheter for congenital diaphragmatic hernia¹¹ are the most recent foetal intervention being carried out in-utero. Non-invasive prenatal diagnosis of foetal RhD status¹² and foetal anaemia has become a reality.¹³

On the other hand, assessment of foetal growth and subsequent monitoring of foetal oxygenation status can be easily performed at the level of the foetal and umbilical vessels and haemodynamic changes can be observed and used for improving the characteristics of management.¹⁴

With these advances, incorporation of the knowledge of genomics, proteomics, and stem cell research and gene therapy into foetal medicine is now required so as to make a wider range of therapeutic interventions possible. Further, the field needs to move into the next phase that is the evidence based for the practice of foetal medicine and surgery where it is important not to introduce exciting techniques without solid evidence which eventually would require establishment of and collaborations between multicentre networks of excellence globally; this would help prioritize and set the agenda for good quality research and clinical progress in this field.

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