Maternal distress: congenital cardiac defects on the morphological scan

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GP Emergency Management articles use real cases to illustrate the emergency management of patients presenting in general practice with cardiac problems.

A pregnant 25-year-old woman comes straight from the radiologist in tears, requesting an urgent appointment. She has just had the 18 to 20-week morphological ultrasound scan and the radiologist told her that the baby's heart is not normal. She says the radiologist is going to ring the practice.

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This patient is usually seen by your colleague who is away and she wants an appointment today. What information would you like to know from her previous records?

Answer: You check her records to see if the patient was seen for preconception advice. Several months ago during a consultation with your colleague, this woman said she wanted to fall pregnant and she was advised to start taking folic acid 400 µg/day. After her pregnancy was confirmed, antenatal blood tests were organised and she was advised to have a nuchal translucency scan at 11 to 14 weeks' gestation. She elected not to have the scan because she was under the misconception that it only gave information about the risk of the baby having Down syndrome. As she is young and had a relatively low risk of Down syndrome (and preferred not to pay for the test), she chose to save the money and only have the 18 to 20-week morphological scan.

An increased nuchal translucency measurement is an indication of much more than just an increased risk of Down syndrome. Nuchal translucency may be increased not only in the presence of a chromosome abnormality but also in the presence of a genetic syndrome or structural abnormality, with cardiac defects being the most common. If performed by an experienced ultrasonographer, the 12-week scan may also detect many major organ abnormalities (such as anencephaly and omphalocoele, and even spina bifida). This allows for earlier interventions and treatment as required. Thankfully, this patient's 18 to 20-week scan was performed by a centre experienced in obstetric imaging with modern equipment.

What do you say and do next?

Answer: You see this patient as soon as possible. This is an emotional emergency and she is likely to be in psychological shock. What you tell her now may not be remembered and sensitively phrasing your sentences is important. Tell her it is no one's fault that this has happened. Reassure her that there is a wide range of ways in which the developing heart can have problems and that many of these problems are manageable. Tell her you want to speak with the radiologist and that you will ring the radiology clinic now. Ask her if she has spoken to her husband.

The radiologist tells you that the fetus was about 18 weeks' gestation from the scan and the dates were approximately 19 weeks. The heart was well-demonstrated and the abnormality is thought to be tetralogy of Fallot (Figure). There appeared to be no other abnormalities.

How should you now manage the situation?

Answer: Explain to the woman that the radiologist believes her baby has a common heart problem that can affect a developing baby. However, as the radiologist is not a specialist in heart abnormalities seen on ultrasound in babies, you will arrange for an urgent appointment with a specialist radiologist. It is also important for the pregnancy to be reviewed by a specialist in fetal medicine, who will clarify the nature of the abnormality and discuss with the patient whether any additional tests need to be performed. It is also likely that a paediatric cardiologist will be involved in advising her and her husband about what the next step might be.

The patient wants to know what the abnormality actually is. What do you tell her?

Answer: Tell her that it is probably, but not definitely, tetralogy of Fallot. This condition involves four heart defects: narrowing of the artery that takes the blood to the lungs from the heart; thickening of the muscle on the right side of the heart (to better push the blood to the lungs through the narrow artery); a connecting hole between the two sides of the heart that normally is not there; and an enlargement of a major artery that straddles both sides of the heart instead of just the left. You tell her there is a lot of variation in the severity of this

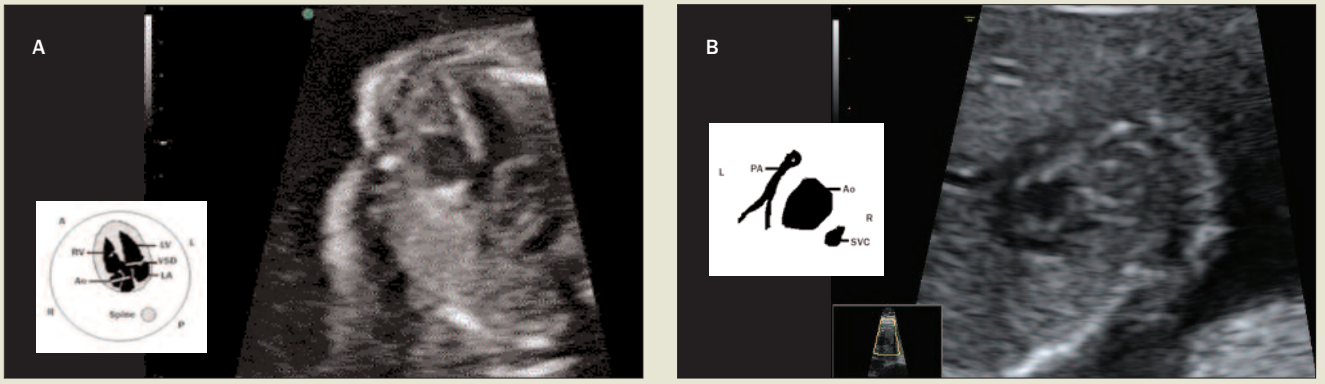

Figure. Fetal echocardiogram showing the intrauterine features of tetralogy of Fallot at 20 weeks' gestation. a (left). Five-chamber view showing large malalignment ventricular septal defect (VSD) with overriding aorta (Ao). b (right). Three-vessel view showing the small pulmonary artery (PA), wide aorta (Ao) and superior vena cava (SVC). Right ventricular hypertrophy is not a feature of in utero tetralogy of Fallot. A = anterior; L = left; LA = left atrium; LV = left ventricle; P = posterior; R = right; RV = right ventricle.

type of abnormality and some babies are not as sick as others. The main problem is that these babies have less oxygen in their blood and therefore look blue rather than pink. All affected babies require corrective cardiac surgery at some stage in the first year of life, depending on how they are managing. A paediatric cardiothoracic surgeon performs this surgery and the success rate is high.

The woman wants to know if she should end the pregnancy. She does not think that she and her husband could cope with a chronically ill child that will die young and it is not fair on the child either. What do you say now?

Answer: Tell the patient that ending the pregnancy is something that she should discuss with the specialists when they have more details about the abnormalities and can explain the consequences. A decision to end a pregnancy should not be taken lightly or be rushed, and is the personal choice of the parents. Ultimately, it is her and her husband's decision to make once they have considered all the information available.

Explain to the patient that this is a common situation and serious heart defects occur in about one in 100 pregnancies.[1] There is a wide range in the severity of the abnormalities found in cases such as hers, and at this stage it is impossible for her to think about what she and her husband should do. If tetralogy of Fallot is present, the baby would certainly need heart surgery, usually at about 6 months of age, depending on how the baby copes with the defect.

As tetralogy of Fallot is the most common of the heart defects found in newborns where the baby looks blue, paediatric cardiothoracic surgeons have a lot of experience with this operation. The survival chances are very good, especially as there are no other structural abnormalities found in this patient's baby.

The woman now wants you to speak to her husband on the telephone because she does not think she will remember all the details. You explain the situation to him and he leaves work to come to your practice. What do you do in the meantime?

Answer: The quality of the ultrasound scans and the experience of the reporting radiologist in fetal medicine are the most important factors in clarifying the situation. You arrange an urgent appointment (ideally the same week) for a repeat morphological ultrasound. Usually such an urgent appointment can be arranged through a tertiary obstetric unit or hospital fetal medicine centre. Ask to speak to a specialist in fetal medicine. Sometimes specialists in fetal medicine work privately (typically with very up-to-date scanning equipment) and this may be a quicker option for the patient.

While you are waiting for the husband, the woman asks you why this might have happened to her. What do you say?

Answer: You reiterate that it is no one's fault and explain that heart defects come about in three main ways. Often a fault in the development occurs without any apparent reason. Another possibility is that the fetus was exposed to something damaging in early pregnancy, such as a drug or perhaps a virus. Finally, heart defects are more likely to occur in genetically related family members of those who have had heart defects.

The patient tells you that she had a hole in the heart that closed without surgery in middle childhood. Her mother has told her since that she would never have any problems with her heart because the hole closed completely.

Why is the patient's heart defect relevant?

Answer: Congenital heart defects are among the most common congenital malformations, with a reported incidence of eight to 10 per 1000 live births.[1] About one-third of defects are severe and are responsible for significant mortality and morbidity in the neonatal period and infancy. There is a 2 to 5% risk of the baby having a heart abnormality if the mother has had a congenital heart defect, although this risk varies depending on the maternal diagnosis. If two or more first-degree relatives are affected by heart defects, there is an even greater risk for the fetus; however, the degree or type of abnormality is not directly hereditary.

Are there any other factors that should be considered if the pregnancy is to continue?

Answer: Amniocentesis may be performed. The fluid should be sent for karyotyping studies and fluorescent cytogenetic testing (fluorescent in situ hybridisation [FISH]). This is a good idea because heart defects may be associated with chromosome defects and genetic syndromes, and this is especially true of tetralogy of Fallot.

Knowledge of these genetic defects may assist this patient and her husband in their decision as to whether to continue with the pregnancy. About 15 to 20% of cases of tetralogy of Fallot will have a 22q11 chromosomal deletion (velo-cardiac-facial syndrome or DiGeorge syndrome) and 8 to 10% will have Down Syndrome (usually in association with an atrioventricular septal defect).[2]

In this case, a review of the morphological scan is important to exclude any other abnormalities in organ development because these may worsen the prognosis.

At some stage, a referral to a clinical geneticist may be useful to discuss recurrence rates and the results of the amniocentesis and its implications, especially if the 22q11 chromosomal deletion is found. Parental DNA analysis may be indicated because one of the parents may also have this deletion.

This couple is enduring an emotional roller coaster and needs to assimilate much information in a relatively short time. The tertiary referral centre will put them in contact with an experienced counsellor. In the meantime, several appointments with you may be necessary to clarify some of the information the couple have been given.

The parents may wish to speak to other parents of children with tetralogy of Fallot so that they can get an idea of what may lie ahead of them. They should be put in contact with a parents' support group such as HeartKids (www.heartkids.org.au).

What is 22q11 deletion syndrome?

Answer: 22q11 deletion syndrome is a genetic syndrome that can be hereditary and occurs in one in 2000 babies. It is the most common cause of mental retardation with congenital heart disease in humans.

22q11 deletion syndrome has a wide range in severity. Most cases are in the milder spectrum and may be easily missed if not suspected and tested for genetically.

The syndrome may be associated with:

- craniofacial abnormalities – cleft palate, a long face that is asymmetrical, missing teeth, small teeth and enamel hypoplasia, microcephaly, cataracts, narrow palpable fissures, strabismus, micro-ophthalmia, mild hypertelorism, puffy upper eyelids, protuberant small cup-shaped ears, mild deafness, narrow nasal passages
- cardiac defects – tetralogy of Fallot (or any of its components in isolation), truncus arteriosus, patent ductus arteriosus, aortic coarctation, valvular abnormalities, vascular abnormalities of major arteries and veins
- neurological abnormalities – periventricular cerebral cysts, cerebellar mental, retardation, dysgenesis, hypotonia, strokes, seizures, spina bifida, psychosis
- genitourinary abnormalities – dysplastic, cystic kidneys, vesico-ureteric reflux, hypospadias, cryptorchidism
- endocrine abnormalities – hypoparathyroidism, hypothyroidism) –
- musculoskeletal problems – orthopaedic deformities, hernias
- immune problems. **CT**

Key points

- **Always refer pregnant patients for nuchal translucency and morphological scans to radiologists who specialise in fetal medicine.**
- **The nuchal translucency scan is important, not just because of the early detection of chromosomal disorders but also because it may be abnormal in the presence of significant congenital abnormalities, especially those relating to the heart.**
- **The diagnosis of a fetal cardiac abnormality is an emotional emergency for the couple concerned and requires empathetic counselling and often referral to a psychosocial specialist.**
- **If fetal cardiac defects are noted, the patient needs an urgent assessment by a fetal medicine specialist with specific expertise in cardiology.**
- **There is a wide variation of abnormalities within the same congenital cardiac syndrome or condition, and each case should be approached individually.**
- **Always ask about a family history of congenital defects (especially cardiac), ideally at the preconception consultation but otherwise as soon as the pregnancy is confirmed.**
- **22q11 deletion syndrome is one of the most common genetic conditions that are hereditary. It occurs in a wide range of clinical variations and may be underdiagnosed.**

References

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2. Roos-Hesselink JW, Kerstjens-Frederikse WS, Meijboom FJ, et al. Inheritance of congenital heart disease. Neth Heart J 2005: 13 (3); 88-91.