

Caesarean section should not be routinely performed. However, if the aortic root is enlarged beyond 4.5cm or there is significant aortic regurgitation, delivery at 39 weeks by induction or caesarean section should be considered, to minimise the effect of high hormone levels on the aortic wall.

Anaesthesia

Epidural is preferable to general anaesthesia, as intubation may lead to marked fluctuations in blood pressure.

Bear in mind the possibility of dural ectasia. Intubation may be difficult due to high palate, limited neck extension, jaw dislocation and floppy larynx.

Genetic Testing

Prenatal diagnosis is now technically possible by genetic linkage, or directly if a mutation has been detected in the affected parent prior to the foetus reaching 11 weeks gestation. A chorionic villus biopsy will then allow mutation screening in the unborn child.

Preimplantation genetic diagnosis is available on a private basis, sometimes with NHS funding through the Primary Care Trust. Eventually this may be made available through the NHS. This process allows the couple to plan to have an unaffected child.

Useful References

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Pregnancy in Marfan Syndrome

INFORMATION FOR OBSTETRICIANS

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What is Marfan Syndrome?

An inherited disorder of connective tissue that affects many organ systems including the skeleton, lungs, eyes, heart and blood vessels.

This condition can affect both men and women of any race or ethnic group. It is estimated that around 18,000 people in the United Kingdom have Marfan syndrome.

Salient Features

Skeletal

Tall thin physique, with long limbs and fingers, scoliosis, narrow chest with pigeon or funnel deformity, joint hypermobility and dislocations. Dural ectasia occurs in 60% of patients.

Cardiovascular

Dilatation of ascending (and sometimes descending) aorta, incompetence of aortic and mitral valves, aneurysm and dissection of aorta.

Respiratory

Pneumothorax, bronchiectasis, emphysema and asthma.

Ocular

Subluxation or dislocation of lens, myopia and unstable refraction, detachment of retina, strabismus, glaucoma.

Dental

High arched palate, crowding of teeth.

Genetic

Males and females are affected equally frequently. Each child of an affected parent has a 50% chance of inheriting Marfan syndrome. In 25% of cases neither parent is affected; however, apparently unaffected parents should be screened carefully as the severity and pattern of disease are variable, even within one family.

Diagnosis

Diagnosis is made after careful physical examination and echocardiography, demonstrating classical features in 2 out of 3 major systems (eyes, heart, skeleton), preferably with a family history.

Diagnosis can be confirmed within a family by genetic linkage studies.

Mutations can be found in the fibrillin-1 gene in 92% of patients, assisting with screening of family members, including prenatal diagnosis.

Cardiac Problems

The most serious problems occur in the heart and blood vessels. The aorta is usually wider than expected for a given body surface area and is more fragile due to a deficiency in the amount of fibrillin present. The dilatation tends to be progressive leading to aortic dissection +/- aortic regurgitation. Surgical repair is recommended when the aortic root becomes widened to 5cm, or earlier in cases with a poor family history of early dissection. Beta-blocker therapy can delay dilatation. Mitral valve prolapse is also often present. Antibiotic prophylaxis is recommended during delivery.

Obstetric Aspects

Obstetric care for the pregnant Marfan patient should be provided by practitioners conversant with the possible complications of the syndrome, and with access to facilities for cardiac surgery should emergencies arise. Counselling by an expert prior to becoming pregnant is an essential step, with special attention paid to the risks of the mother, the 50% risk to the child of inheriting the syndrome and the necessity for optimising the mother's medical condition prior to conception. The degree to which any child will be affected can be gauged largely by observing the natural history in other family members.

The risk to the mother of major complications such

as aortic dilatation, aortic rupture and increasing mitral or aortic regurgitation should be considered. Preconceptual dilatation of the aorta beyond 4cm has been suggested in 2 studies to be an important but not foolproof predictor of complications during gestation. If the aorta, at its widest, is greater than 4cm or there is moderate to severe aortic or mitral regurgitation, then pregnancy is particularly risky. Dilatation of the aorta beyond 4cm is also the point at which cardio-selective beta-blockers should be introduced and maintained throughout pregnancy.

The overall risk of a sudden serious complication is in the order of 5-10% if aortic root diameter is over 4cm with aortic regurgitation. Aortic root replacement with preservation of aortic valve should be offered if aortic root diameter is greater than 4.3cm, and pregnancy planned 1 year postoperatively, to reduce risk of aortic dissection in pregnancy.

The most reliable method of predicting the major cardiac complications is with the use of echocardiography.

Echocardiography should be performed 6-8 weeks throughout pregnancy and during the initial 6 month post-partum period.

Blood pressure should be regularly monitored and hypertension treated aggressively.

Other less serious complications to bear in mind include increased joint pain, respiratory complications secondary to scoliosis and widespread abdominal striae. The need for close monitoring of the pregnancy may entail frequent visits to a hospital with specialist facilities and this should be borne in mind by the prospective mother. Near the time of delivery it is unwise for the mother to travel far from the specialist centre.

Delivery should be by the least stressful method; ideally a vaginal delivery in the lateral decubitus position with adequate oxygenation and minimal maternal expulsive efforts. Liberal use of forceps or vacuum extraction is advised to shorten the second stage of labour.