Thrombocytopenia with absent radius syndrome (TAR)

Patient Information Series – What you should know, what you should ask.

What is TAR syndrome?

Thrombocytopenia Absent Radius (TAR) Syndrome is a rare genetic condition characterised by low platelet count (blood cells that help stop bleeding) and absence of a bone called radius in both forearms.

How does a TAR syndrome happen?

TAR syndrome affects fewer than 1 in 100 000 people. It is autosomal recessive condition which means that the syndrome is caused by the abnormal gene inherited from each of the parents. The affected gene is called RBM8A and it is usually paired with deletion affecting chromosome 1.

Should I have more tests done?

When TAR syndrome is suspected before the baby is born, there are three tests that can be offered to you to confirm the condition:

- **Chorionic villi sampling** – taking a sample of the placenta by passing a thin needle through your abdomen and the womb into the placenta and performing genetic testing on the sample.
- **Amniocentesis** – taking a small amount of amniotic fluid by passing a thin needle through the abdominal wall into the womb and the fluid surrounding the baby. The amniotic fluid contains the baby's skin cells that are then used for genetic testing.
- **Cordocentesis** – taking the baby's blood sample from the cord by inserting a thin needle through your abdomen and wall of the womb into the cord. This test is performed to check the baby's platelet count. TAR syndrome can be confirmed if the platelet count is reduced and there are typical arm abnormalities present on scan.

What are the things to watch for during pregnancy?

Regular scans should be performed to exclude any associated abnormalities and to monitor for any signs of bleeding, particularly on the baby's brain. The most frequent associated abnormalities are kidney and heart defects. You may be advised to have a caesarean section instead of vaginal birth as that would reduce the risk of bleeding in the baby's brain at birth.

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What does it mean for my baby after it is born?

The main concern after the baby is born is the risk of bleeding (haemorrhage) that can be severe and life-threatening. The risk is particularly high in the first year of life after which it gradually reduced as the platelet count increases to normal levels in adulthood.

The mildest manifestation of low platelet count are nosebleeds and easy bruising. The most severe complications are bleeds in the brain and other internal organs. Intellectual development can be affected in children who have brain damage due to a haemorrhage. The risk of bleeding can be reduced by platelet transfusions.

Cow's milk allergy is a common symptom in people with TAR syndrome and can worsen the platelet count. Therefore, it is recommended to avoid cow's milk in the diet.
The other less concerning issue is the abnormality of the arms (and sometimes legs) that might require surgery. The other issues will occur if there are any additional abnormalities found before or after birth.

Will it happen again?

If your child was affected by TAR syndrome genetic counselling is recommended. Both you and your partner will be tested for the faulty genes and if you are confirmed as carriers there is a 1 in 4 chance of having another baby with TAR syndrome. Unfortunately, there is nothing that can be done to prevent this condition.

What other questions should I ask?

- How often will I have ultrasound examinations done?
- What options do I have before the baby is born?
- Is there anything that can be done for the baby before birth?
- How and where should I give birth?
- Can I meet the team of doctors that will be assisting my baby when it is born in advance?
- Will my baby need surgery after birth?

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