

TAR Syndrome

BIRTH DEFECT RESEARCH FOR CHILDREN



What is TAR Syndrome?

Thrombocytopenia Absent Radius Syndrome (TAR)



TAR Syndrome



Thrombocytopenia Absent Radius (TAR) Syndrome is a rare genetic disorder. It is characterized by low levels of platelets in the blood (thrombocytopenia), absence (aplasia) of the bone on the thumb side of the forearm (radius) on both arms, and underdevelopment (hypoplasia) or absence of the bone on the pinky-side of the forearm (ulna). Platelets are very important for normal blood clotting. Consequently, thrombocytopenia results in potentially severe bleeding episodes (hemorrhaging), primarily during infancy. Children with TAR Syndrome frequently have malformations of their hands, but their thumbs are always present. Other abnormalities may include malformations of the heart, legs, and hip sockets; kidney defects; and mental retardation resulting from bleeding in the skull.

How many children have TAR Syndrome?

TAR Syndrome is very rare. There are over 100 known cases, and it has occurred slightly more often in girls. It has not been identified with a specific geographic location or ethnic group.

How do you know if your child has TAR Syndrome?

The characteristic skeletal abnormalities of TAR Syndrome can be detected prenatally by ultrasound from the 16th week. Otherwise, these abnormalities are apparent at birth. Blood tests can reveal the low platelet counts associated with thrombocytopenia.

What causes TAR Syndrome?

Although environmental factors cannot be completely ruled out, it is generally considered an autosomal recessive syndrome, which means that the gene is carried by both parents. There is then a one in four chance of an affected child in each pregnancy. Genetic counseling is recommended after an affected child is identified. The defects occur between 4-8 weeks of fetal development. No ethnic or racial predisposition has been identified and there is no known prevention.

How can you help a child with TAR Syndrome?

Careful handling of your infant, crib padding, and soft helmets can help reduce chances of injury, bruising, and bleeding. Medication can also be used to decrease bleeding. It is important to avoid stress, infections, and surgery during your child's first year since they may precipitate thrombocytopenia. You should eliminate cow's milk from your child's diet if intolerance is identified. Corrective orthopedic braces may be necessary for your child's forearms along with occupational therapy. Orthopedic surgery may be performed once your child has outgrown the severe danger of thrombocytopenia. Platelet or whole blood transfusions may be necessary in cases where there is a severe risk of bleeding. Internal bleeding requires immediate medical care. Consult your doctor if your child suddenly bruises more frequently, develops unexpected and prolonged bleeding, develops a rash of red pin-prick spots, is drowsy or unresponsive, or seems to be experiencing a lot of pain. A team of medical specialists is recommended for children with TAR Syndrome and usually should include an orthopedic surgeon, plastic surgeon, hematologist, and cardiologist.

What's in the future for a child with TAR Syndrome?

Children with TAR Syndrome experience frequent bleeding because of the thrombocytopenia. Thirty-five to forty percent of them die from bleeding in the brain, almost all before their first birthday. For those who survive intracranial bleeding, mental retardation is possible. Children commonly experience delayed motor development due to the hand deformities. The low platelet counts improve to

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almost normal by adulthood. The prognosis appears good for those children who survive the first two years. Most have a normal life span and are able to live independently and have productive jobs. Many adults have married and have unaffected children.

Fact Sheet by:

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