



My46 Trait Profile

Diamond Blackfan anemia

Diamond Blackfan anemia is a rare genetic condition associated with anemia. Individuals with Diamond Blackfan anemia may be born with certain physical findings and may be at increased risk to develop blood cancers or tumors. It is caused by mutations in one of nine ribosome genes that are necessary for our body to produce proteins.

Characteristics of Diamond Blackfan anemia

Anemia occurs when the bone marrow does not make enough red blood cells. Red blood cells carry oxygen throughout our bodies. If our body does not have enough red blood cells, our organs do not get enough oxygen. Diamond Blackfan anemia (DBA) is a rare genetic condition characterized by anemia, physical abnormalities, and an increased risk for certain types of cancer.

The majority of children with DBA develop anemia in the first few months of life. About half of children with DBA are born with a noticeable physical finding or difference, such as a cleft lip/palate, unusual thumb shape, missing kidney, or a heart defect. DBA is associated with an increased risk of developing blood cancers, such as acute myelogenous leukemia (AML) or myelodysplastic syndrome (MDS), and solid tumors, such as osteosarcoma (bone cancer).

Diagnosis/Testing

Nine genes have been identified to cause DBA: RPS19, RPL5, RPS10, RPL11, PRL35A, RPS26, RPS24, RPS17, RPS7, RPL26, and GATA1. Each of these genes encodes the cell's ribosomes, which are important in the process of protein production. Proteins are the functional units within our cells. Genetic changes or mutations in any one of these genes are thought to interfere with this process, resulting in protein production problems. Approximately 60% of individuals with DBA will have an identifiable mutation in one of these genes.

Management/Surveillance

DBA is treated with blood transfusions, steroid medications, and bone marrow transplant. Blood transfusions every few weeks provide healthy red blood cells to carry oxygen. Certain steroid medications (corticosteroids) are also prescribed. The majority of children with DBA respond to steroid treatment. Bone marrow transplant is the only way to cure anemia symptoms. Bone marrow cells from a healthy donor are used to replace the unhealthy bone marrow cells in patients with DBA.

Mode of inheritance

Autosomal dominant inheritance:

In most cases, DBA is inherited in an autosomal dominant pattern. This means inheriting one gene mutation is enough for an individual to be affected and show signs of DBA. The mutation can be inherited from a parent, however almost half of individuals with DBA have a new (de novo) mutation.

X-linked recessive inheritance:

Mutations in the GATA1 gene cause X-linked recessive DBA. The gene mutations causing this type of inheritance is found on the X chromosome. An X-linked recessive pattern means that in females, both copies of the GATA1 gene (i.e., one on each X chromosome) must have a mutation, whereas in males, only one copy of the GATA1 gene must

have a mutation to be affected. A female with a mutation in one copy of the GATA1 gene on the X chromosome is said to be a carrier for an X-linked condition, and is typically not affected.

Risk to family members

Autosomal dominant inheritance:

The risk to family members depends on whether or not the individual with DBA has a parent affected with DBA. If a parent also has the condition, the risk of having a child with DCM is 50% with each pregnancy. If a parent does not have DCM, the risk of other siblings being affected is very low.

X-linked recessive inheritance:

If a father is affected with X-linked recessive DBA, his daughters will be carriers of DBA and his sons will be unaffected. If a mother is a carrier of DBA, each daughter has a 1 in 2 chance (i.e., 50%) of being a carrier and each son has a 1 in 2 chance (i.e., 50%) of being affected with DBA.

Special considerations

None

Resources

Diamond Blackfan Anemia Foundation, Inc.

<http://dbafoundation.org>

Genetics Home Reference: Diamond-Blackfan anemia

<http://ghr.nlm.nih.gov/condition/diamond-blackfan-anemia>

References

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