Fanconi Anemia: Type A (FANCA)

Fanconi Anemia can be caused by defects in a number of genes, one of which is FANCA. This gene is involved in producing a protein involved in DNA repair, which it carries out via the so-called Fanconi anemia pathway. If the protein fails to function, DNA repair will not be carried out as normal, which can lead to many abnormalities, particularly affecting the bone marrow and blood cells. Patients have anemia and tend to suffer from infections. They are much more at risk of leukemia and other cancers than the general population. The majority of patients have one or more physical abnormalities, although a large minority are physically normal. A wide range of physical problems are possible. Common issues include short statue, unusual skin pigmentation, misshapen thumbs, microcephaly, eye defects, and deformed kidneys or genitals. The majority of those with the disease die before the age of 30.

The overall incidence of Fanconi anemia is roughly 1 in 160,000, of which about 60-70% are due to defects in the FANCA gene. Some populations, such as Spanish Roma, black South Africans, and Ashkenazi Jews, are at greater risk of the disease. Fanconi anemia type A is inherited in an autosomal recessive manner, typically requiring both parents to carry a faulty gene asymptomatically.

Sources

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