Bloom Syndrome (BLM)

Bloom syndrome is caused by mutations in the BLM gene, which encodes for one of the RecQ helicase proteins. These proteins have an important role in preserving the integrity of DNA, as well as catalyzing key reactions that are crucial for DNA unwinding. Those with the disease have unusually short stature, and are very sensitive to sunlight, often having reddish marks on their faces. Men are sterile, while women have reduced fertility with an early onset of menopause. Most sufferers are of normal intellectual ability, although some suffer from learning difficulties. Cancer is much more likely in those with Bloom syndrome, often first appearing in their 20s or 30s. Early mortality from cancer is common, although sufferers often respond successfully to treatment.

Bloom disease is an extremely rare condition, with about 300 cases known worldwide, about a quarter of which are among those Ashkenazi Jewish descent. The condition is autosomal recessive, which typically requires an affected child to have two asymptomatic carrier parents.

Sources

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