DELETION 14Q23

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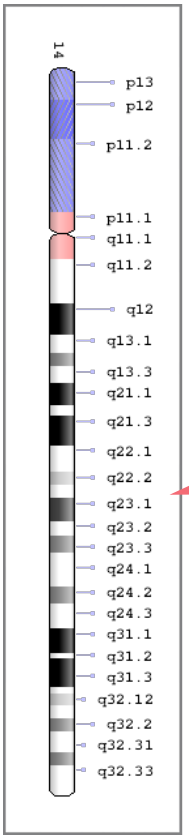

Deletions of the segment q23 on chromosome 14 occur rarely, but their clinical manifestations are very unusual. This area contains the SPTB gene which has been associated with spherocytosis. Specifically, SPTB encodes beta-spectrin, which is an incredibly important cell membrane protein in red blood cells[1]. As a result of this defect, red blood cells which normally have a disk-like structure became mostly spheric. This condition is called spherocytosis. Red blood cells in such patients have a decreased ability to transfer oxygen and more likely to be destroyed (hemolyzed) that leads to anemia.

Spherocytosis is the main but not only manifestation of the syndrome of 14q23 deletion. Most patients also reveal neurological findings while some have also documented physical findings and congenital heart defects.

Spherocytosis, a frequent finding in patients with 14q23 deletions, is the most common nonimmune hemolytic anemia, affecting anywhere between 1:2000 to 1:5000 caucasians[1]. Typically inherited in an autosomal dominant manner[2], the disease itself is caused by a disruption to the vertical links that fix a red blood cell's phospholipid bilayer to its cytoskeleton[1]. Individuals with this disease can manifest several symptoms that can vary in severity from almost no symptoms to severe hemolysis, splenomegaly, and gallstones[1]. Jaundice has also been noted in some patients[2].

Patients with 14q23 deletions may also exhibit other neurological findings. This can include things like developmental delay, epilepsy,

or hypoplasia of the corpus callosum[3]. Additionally, some individuals may also have various features of an autism spectrum disorder diagnosis[4] or motor discoordination[1].

Although spherocytosis and neurological findings are the most common, there are some physical features that have been noted in various studies. Up-slanting palpebral fissures, supraorbital lateral fullness, thickened ears, macrocephaly, and obesity have been documented[4]. There has also been documentation of some congenital heart defects including ventricular septal defect and bicuspid aortic valve[3].

REFERENCES

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