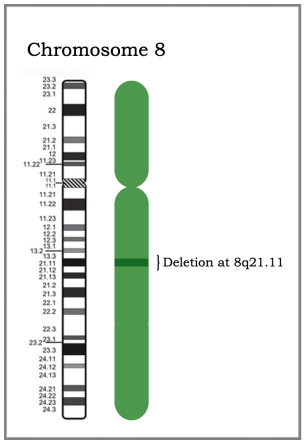
YOU ARE NOT ALONE

CHROMOSOME DISORDER OUTREACH

8Q21.11 DELETION

8q21.11 Deletion



Individuals with deletions on the long arm of chromosome 8 encompassing the q21.11 region are frequently characterized by intellectual disability, motor delays, hypotonia, distinct facial features, ocular abnormalities and defects of the hands and feet[1]. The extent of these features, however, can vary from individual to individual.

It is common for individuals with 8q21.11 deletions to experience mild to moderate intellectual disability, although global developmental delays may also be seen. Some children with 8q21.11 deletions may be several months behind in achieving speech, motor, and personal-social skill milestones[2]. Many of the gross motor delays that are seen is due to hypotonia of the lower extremities. As a result, some individuals may benefit from physical, occupational, or mobility therapy[3].

There are several distinct facial features frequently identified in those with 8q21.11 deletions. This includes having a round or square face with a high forehead, downslanting palpebral fissures, ptosis, wide nasal bridge, cupid's bow or tented upper lip, high-arched palate, micrognathia, and low set ears[1].

In addition to these common facial features, specific ocular findings are commonly seen. A specific finding, called Peters anomaly, is frequently seen. Peters anomaly is a type of anterior segment dysgenesis of the eyes that is characterized by cloudiness of the cornea due an incomplete separation of the lens and cornea during early development[3]. This opacity often results in blurry vision and may

require surgical treatment[4]. Additionally, strabismus and other refractory abnormalities can also be seen[1].

Individuals with 8q21.11 deletions frequently have abnormalities of the hands and feet. Specifically, syndactyly, camptodactyly, brachydactyly, and abnormal palmar creases are typically seen[1].

While less common, some patients may present with heart defects. This includes atrial septal defects, patent ductus arteriosus and cardiomyopathy[2].

There are a few genes that are thought to play crucial roles in the development of the features typically seen in those with 8q21.11 deletions. ZFHX4 is known to act as a transcription factor that plays a role in neural cell development. It is thought that deletion of this gene may contribute to the intellectual disability and hypotonia seen in those with 8q21 deletions[3]. There is mixed evidence if ZFHX4 plays a role in ocular development as well[1,3]. Other genes, such as PEX2 and HEY1, are suggested to also play significant roles in the characteristic features of 8q21.11 deletions, though more data is needed to define their role[2,5].

REFERENCES

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