A Rare Aarskog Genetic Syndrome

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Introduction

Aarskog-Scott syndrome is a hereditary condition that affects how different body parts develop. Although some females may exhibit modest symptoms of the disease, men are primarily affected by this condition. Short stature and numerous facial, limb, and genital deformities are hallmarks of the rare genetic disorder known as Aarskog syndrome. In some cases, certain forms of cognitive impairments may also exist. The only gene known to be connected to Aarskog syndrome is the FGD1 gene on the X chromosome. It is thought that Aarskog-Scott syndrome is a rare disease. Since persons who are only minimally affected may not receive a diagnosis, its incidence is unknown. Aarskog-Scott syndrome is known to have a hereditary aetiology that involves mutations in the FGD1 gene. Instructions for creating a protein that activates another gene are provided by the FGD1 gene [1].

Description

Males are the main victims of Aarskog syndrome. Boys who are affected display a distinctive set of genital, skeletal and facial anomalies. Even within families, clinical symptoms can differ from person to person. Males with Aarskog syndrome have a large forehead and rounded face. Ocular hypertelorism, ptosis, palpebral fissures, a tiny nose with flared forward nostrils, maxillary hypoplasia and a widow's peak are other distinguishing facial characteristics. Female carriers frequently have a few mild signs of the condition in their hands and cheeks. A rare malformation of the external genitalia called penoscrotal transposition causes the scrotum to be positioned incorrectly over the penis. A protein with atypical function is produced as a result of mutations in the FGD1 gene.

Faciogenital dysplasia (FGD, also known as Aarskog-Scott syndrome (AAS), is an X-linked condition with a recessive mode of inheritance. A distinctive combination of short height, genital, facial and skeletal deformities define this disorder. The latter include small bird, short nose, syndactylyy and hypertelorism. Ploiss, joint hyperexstensibility and mental retardation are further characteristics. Since patient clinical presentations vary greatly, it is difficult to make a definitive diagnosis. According to genetic and biochemical research, FGD1 encodes a guanine nucleotide exchange factor (GEF), also known as an activator, for the Raslike GTPase Cdc42, a member of the Rho family. Rho protein is expressed in precartilaginous mesenchymal condensations, the perichondrium and periosteum, proliferating chondrocytes and osteoblasts inside the growing mouse skeleton. These findings indicate that mesenchymal prechondrocytes, chondrocytes and osteoblasts are just a few of the skeletal cell types for which FGD1 signalling may be important.

Short stature and numerous facial, limb and genital deformities are hallmarks of the rare genetic disorder known as Aarskog syndrome. In some cases, certain forms of cognitive impairments may also exist. The sole gene connected to Aarskog syndrome is the FGD1 gene on the X chromosome, contains the FGD1 gene. One mutated copy of the gene in each cell is sufficient to induce the disease in males (who have only one X chromosome). To produce Aarskog-Scott syndrome in females (who have two X chromosomes), a mutation would need to exist in both copies of the gene [4].

The FGD1 gene has undergone mutation, which causes the Aarskog-Scott syndrome. Cdc42, a member of the Rho (Ras homology) family of the p21 GTPases, is particularly activated by the GEF that FGD1 encodes. FGD1 protein promotes fibroblasts to generate filopodia, cytoskeletal components involved in cellular communication, adhesion and migration. This is accomplished by activating Cdc42. The c-Jun N-terminal kinase (JNK) signalling cascade, which controls cell proliferation, apoptosis and cellular differentiation, is activated by FGD1 protein via Cdc42 [5].

Conclusion

FGD1 protein is expressed in precartilaginous mesenchymal condensations, the perichondrium and peristome, proliferating chondrocytes and osteoblasts inside the growing mouse skeleton. These findings indicate that mesenchymal prechondrocytes, chondrocytes and osteoblasts are just a few of the skeletal cell types for which FGD1 signalling may be important.

Acknowledgement

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Conflict of Interest

There are no conflicts of interest by author.

References

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