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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 shawl scrotum, small nose, syndactyly and hypertelorism. Ptosis, joint hyperextensibility 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 proteins are a family of at least eight unique proteins that regulate a variety of cellular processes, including the arrangement of the actin filaments.

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 as of yet is the FGD1 gene on the X chromosome. Aarskog-Scott syndrome patients frequently exhibit distinguishing facial characteristics such widow's peak hairlines, tiny noses, lengthy areas between the nose and lips and widely spread eyes (hypertelorism). By childhood, they generally exhibit mild to moderate short stature, but during adolescence, their growth typically catches up with that of their classmates. Short fingers (brachydactyly), curled pinky fingers (fifth finger clinodactyly), webbing of the fingers and other hand deformities are frequent with this syndrome.

Of the 23 pairs of chromosomes that a human has, the last pair is often anasomal, which means that it controls a child's sex. A female has two X chromosomes, whereas male progeny have one X and one Y chromosome. This particular condition is recessive and X-linked. Therefore, if a mutation occurs in a male with only one X-chromosome, the condition will probably manifest itself. If one of the two X chromosomes in a female is mutated, the other X chromosome compensates and she has the syndrome; nonetheless, there are no significant implications. Such folks never show any symptoms of the illness [2,3].

Aarskog-Scott syndrome is a hereditary condition that mostly affects the development of the head and face, hands and feet, genitalia and urinary system (genitourinary tract). Although some females may exhibit modest symptoms of the disease, men are primarily affected by this condition. Aarskog-Scott syndrome is inherited in an X-linked recessive form when it is brought on by FGD1 gene mutations. One of the two sex chromosomes, 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 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.

Acknowledgement

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Conflict of Interest

There are no conflicts of interest by author.

References

1. Rajkumar, S.V., V. Gupta, R. Fonseca and A. Dispenzieri, et al. "Impact of primary molecular cytogenetic abnormalities and risk of progression in smoldering multiple myeloma." *Leukemia* 27 (2013): 1738-1744.
2. Qiu, Li-Mei, Wen-Jian Li, Xin-Yue Pang and Qing-Xiang Gao, et al. "Observation of DNA damage of human hepatoma cells irradiated by heavy ions using comet assay." *World J Gastroenterol WJG* 9 (2003): 1450.
3. Bergsagel, P. Leif, Marta Chesi, Elena Nardini and Leslie A. Brents, et al. "Promiscuous translocations into immunoglobulin heavy chain switch regions in multiple myeloma." Proc Natl Acad Sci 93 (1996): 13931-13936.
4. Fonseca, Rafael, Peter Leif Bergsagel, Johannes Drach and John Shaughnessy, et al. "International Myeloma Working Group molecular classification of multiple myeloma: spotlight review." *Leukemia* 23 (2009): 2210-2221.
5. Bergsagel, P. Leif, María-Victoria Mateos, Norma C. Gutierrez and S. Vincent Rajkumar, et al. "Improving overall survival and overcoming adverse prognosis in

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the treatment of cytogenetically high-risk multiple myeloma." *Am J Hematol* 121 (2013): 884-892.

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