Jackson-Weiss Syndrome

Subjects: Genetics & Heredity

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Jackson-Weiss syndrome is a genetic disorder characterized by foot abnormalities and the premature fusion of certain skull bones (craniosynostosis).

genetic conditions

1. Introduction

This early fusion prevents the skull from growing normally and affects the shape of the head and face.

Many of the characteristic facial features of Jackson-Weiss syndrome result from premature fusion of the skull bones. Abnormal growth of these bones leads to a misshapen skull, widely spaced eyes, and a bulging forehead.

Foot abnormalities are the most consistent features of Jackson-Weiss syndrome. The first (big) toes are short and wide, and they bend away from the other toes. Additionally, the bones of some toes may be fused together (syndactyly) or abnormally shaped. The hands are almost always normal.

Some individuals with Jackson-Weiss syndrome have hearing impairment. People with Jackson-Weiss syndrome usually have normal intelligence and a normal life span.

2. Frequency

Jackson-Weiss syndrome is a rare genetic disorder; its incidence is unknown.

3. Causes

Mutations in the *FGFR2* gene cause Jackson-Weiss syndrome. This gene provides instructions for making a protein called fibroblast growth factor receptor 2. Among its multiple functions, this protein signals immature cells to become bone cells during embryonic development. A mutation in a specific part of the *FGFR2* gene overstimulates signaling by the FGFR2 protein, which promotes the premature fusion of skull bones and affects the development of bones in the feet.

3.1. The gene associated with Jackson-Weiss syndrome

- FGFR2

4. Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

5. Other Names for This Condition

- JWS

References

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