Beare-Stevenson Cutis Gyrata Syndrome

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Beare-Stevenson cutis gyrata syndrome is a genetic disorder that typically features skin abnormalities and the premature fusion of certain bones of the skull (craniosynostosis). This early fusion prevents the skull from growing normally and affects the shape of the head and face.

Keywords: genetic conditions

1. Introduction

Many of the characteristic facial features of Beare-Stevenson cutis gyrata syndrome result from the premature fusion of the skull bones. The head is unable to grow normally, which leads to a cloverleaf-shaped skull, wide-set and bulging eyes, ear abnormalities, and an underdeveloped upper jaw. Early fusion of the skull bones also affects the growth of the brain, causing delayed development and intellectual disability.

A skin abnormality called cutis gyrata is also characteristic of this disorder. The skin has a furrowed and wrinkled appearance, particularly on the face, near the ears, and on the palms and soles of the feet. Additionally, thick, dark, velvety areas of skin (acanthosis nigricans) are sometimes found on the hands and feet and in the genital region.

Additional signs and symptoms of Beare-Stevenson cutis gyrata syndrome can include a blockage of the nasal passages (choanal atresia), a malformation of the airways (tracheal cartilaginous sleeve), overgrowth of the umbilical stump (tissue that normally falls off shortly after birth, leaving the belly button), and abnormalities of the genitalia and anus. The medical complications associated with this condition are often life-threatening in infancy or early childhood.

2. Frequency

Beare-Stevenson cutis gyrata syndrome is a rare genetic disorder; its incidence is unknown. Approximately 25 people with this condition have been reported worldwide.

3. Causes

Mutations in the *FGFR2* gene cause Beare-Stevenson cutis gyrata syndrome. This gene produces a protein called fibroblast growth factor receptor 2, which plays an important role in signaling a cell to respond to its environment, perhaps by dividing or maturing. A mutation in the *FGFR2* gene alters the protein and promotes prolonged signaling, which is thought to interfere with skeletal and skin development.

Some individuals with Beare-Stevenson cutis gyrata syndrome do not have identified mutations in the *FGFR2* gene. In these cases, the cause of the condition is unknown.

3.1. The Gene Associated with Beare-Stevenson Cutis Gyrata 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. All reported cases have resulted from new mutations in the gene, and occurred in people with no history of the disorder in their family.

5. Other Names for This Condition

- cutis gyrata syndrome of Beare and Stevenson
- cutis gyrata syndrome of Beare-Stevenson

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