SADDAN

Description

SADDAN (severe achondroplasia with developmental delay and acanthosis nigricans) is a rare disorder of bone growth characterized by skeletal, brain, and skin abnormalities.

All people with this condition have extremely short stature with particularly short arms and legs. Other features include unusual bowing of the leg bones; a small chest with short ribs and curved collar bones; short, broad fingers; and folds of extra skin on the arms and legs. Structural abnormalities of the brain cause seizures, profound developmental delay, and intellectual disability. Several affected individuals also have had episodes in which their breathing slows or stops for short periods (apnea). Acanthosis nigricans, a progressive skin disorder characterized by thick, dark, velvety skin, is another characteristic feature of SADDAN that develops in infancy or early childhood.

Frequency

This disorder is very rare; it has been described in only a small number of individuals worldwide.

Causes

Mutations in the *FGFR3* gene cause SADDAN. The *FGFR3* gene provides instructions for making a protein that is involved in the development and maintenance of bone and brain tissue. A mutation in this gene may cause the FGFR3 protein to be overly active, which leads to the disturbances in bone growth that are characteristic of this disorder. Researchers have not determined how the mutation disrupts brain development or causes acanthosis nigricans.

Learn more about the gene associated with SADDAN

- FGFR3

Inheritance

SADDAN is considered an autosomal dominant disorder because one mutated copy of the *FGFR3* gene in each cell is sufficient to cause the condition. The few described

cases of SADDAN have been caused by new mutations in the *FGFR3* gene and occurred in people with no history of the disorder in their family. No individuals with this disorder are known to have had children; therefore, the disorder has not been passed to the next generation.

Other Names for This Condition

- Achondroplasia, severe, with developmental delay and acanthosis nigricans
- SADDAN dysplasia
- Severe achondroplasia with developmental delay and acanthosis nigricans
- Skeleton-skin-brain syndrome
- SSB syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Severe achondroplasia-developmental delay-acanthosis nigricans syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2674173/)

Genetic and Rare Diseases Information Center

- Severe achondroplasia-developmental delay-acanthosis nigricans syndrome (https://rarediseases.info.nih.gov/diseases/9443/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- ACHONDROPLASIA, SEVERE, WITH DEVELOPMENTAL DELAY AND ACANTHOSIS NIGRICANS; SADDAN (https://omim.org/entry/616482)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28saddan%5BTIAB%5D%29+OR+%28severe+achondroplasia+with+developmental+delay+and+acanthosis+nigricans%5BTIAB%5D%29+OR+%28skeleton-skin-brain+syndrome%5BTIAB%5D%29+OR+%28ssb+syndrome%5BTIAB%5D%29+OR+%28saddan+dysplasia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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