Bowen-Conradi Syndrome

Subjects: Genetics & Heredity
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Bowen-Conradi syndrome is a disorder that affects many parts of the body and is usually fatal in infancy. Affected individuals have a low birth weight, experience feeding problems, and grow very slowly. Their head is unusually small overall (microcephaly), but is longer than expected compared with its width (dolichocephaly). Characteristic facial features include a prominent, high-bridged nose and an unusually small jaw (micrognathia) and chin. Affected individuals typically have pinky fingers that are curved toward or away from the ring finger (fifth finger clinodactyly) or permanently flexed (camptodactyly), feet with soles that are rounded outward (rocker-bottom feet), and restricted joint movement.

Keywords: genetic conditions

1. Introduction

Other features that occur in some affected individuals include seizures; structural abnormalities of the kidneys, heart, brain, or other organs; and an opening in the lip (cleft lip) with or without an opening in the roof of the mouth (cleft palate). Affected males may have the opening of the urethra on the underside of the penis (hypospadias) or undescended testes (cryptorchidism).

Babies with Bowen-Conradi syndrome do not achieve developmental milestones such as smiling or sitting, and they usually do not survive more than 6 months.

2. Frequency

Bowen-Conradi syndrome is common in the Hutterite population in Canada and the United States; it occurs in approximately 1 per 355 newborns in all three Hutterite sects (leuts). A few individuals from outside the Hutterite community with signs and symptoms similar to Bowen-Conradi syndrome have been described in the medical literature. Researchers differ as to whether these individuals have Bowen-Conradi syndrome or a similar but distinct disorder.

3. Causes

Bowen-Conradi syndrome is caused by a mutation in the *EMG1* gene. This gene provides instructions for making a protein that is involved in the production of cellular structures called ribosomes, which process the cell's genetic instructions to create new proteins. Ribosomes are assembled in a cell compartment called the nucleolus.

The particular *EMG1* gene mutation known to cause Bowen-Conradi syndrome is thought to make the protein unstable, resulting in a decrease in the amount of EMG1 protein that is available in the nucleolus. A shortage of this protein in the nucleolus would impair ribosome production, which may reduce cell growth and division (proliferation); however, it is unknown how *EMG1* gene mutations lead to the particular signs and symptoms of Bowen-Conradi syndrome.

3.1. The Gene Associated with Bowen-Conradi Syndrome

- EMG1

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- Bowen Hutterite syndrome
- Bowen syndrome, Hutterite type
- Bowen-Conradi Hutterite syndrome
- BWCNS
- Hutterite syndrome

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