Feingold Syndrome

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Feingold syndrome is a disorder that affects many parts of the body. There are two types of Feingold syndrome, distinguished by their genetic cause; both types have similar features that can vary among affected individuals.

Keywords: genetic conditions

1. Introduction

Individuals with Feingold syndrome type 1 or type 2 have characteristic abnormalities of their fingers and toes. Almost all people with this condition have a specific hand abnormality called brachymesophalangy, which refers to shortening of the second and fifth fingers. Other common abnormalities include fifth fingers that curve inward (clinodactyly), underdeveloped thumbs (thumb hypoplasia), and fusion (syndactyly) of the second and third toes or the fourth and fifth toes.

Additional common features of both types of Feingold syndrome include an unusually small head size (microcephaly), a small jaw (micrognathia), a narrow opening of the eyelids (short palpebral fissures), and mild to moderate learning disabilities. Less often, affected individuals have hearing loss, short stature, or kidney or heart abnormalities.

People with Feingold syndrome type 1 are frequently born with a blockage in part of their digestive system called gastrointestinal atresia. In most cases, the blockage occurs in the esophagus (esophageal atresia) or in part of the small intestine (duodenal atresia). Individuals with type 2 do not have gastrointestinal atresias.

2. Frequency

Feingold syndrome appears to be a rare condition, although its exact prevalence is unknown. Type 1 is more common than type 2.

3. Causes

Mutations in the *MYCN* gene cause Feingold syndrome type 1, and mutations in chromosome 13 that remove (delete) a region of the chromosome that includes the *MIR17HG* gene cause type 2. Both genes are involved in growth and development, particularly before birth.

The *MYCN* gene provides instructions for making a protein that regulates the activity (expression) of other genes. The protein attaches (binds) to specific regions of DNA and controls the first step of protein production (transcription). Studies suggest that the MYCN protein is necessary for normal development of the limbs, heart, kidneys, lungs, nervous system, and digestive system.

The *MIR17HG* gene provides instructions for making a set of microRNAs (miRNAs) known as the miR-17~92 cluster. MiRNAs are short pieces of RNA, a chemical cousin of DNA. These molecules control gene expression by blocking protein production. The miRNAs in the miR-17~92 cluster are involved in the development of many tissues and organs in the body.

Mutations affecting the *MYCN* or *MIR17HG* gene that cause Feingold syndrome prevent one copy of the gene in each cell from producing any functional protein or miRNAs, respectively. As a result, only half the normal amount of the protein or miRNAs is available to control the activity of specific genes during development. It remains unclear how a reduced amount of the MYCN protein or miR-17~92 cluster miRNAs cause the specific features of Feingold syndrome.

3.1. The Genes and Chromosome Associated with Feingold Syndrome

- MIR17HG

- MYCN
- chromosome 13

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

- Brunner-Winter syndrome
- microcephaly-mesobrachyphalangy-tracheoesophageal fistula (MMT) syndrome
- microcephaly-oculo-digito-esophageal-duodenal (MODED) syndrome
- oculo-digito-esophagoduodental (ODED) syndrome

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