Autosomal Recessive Primary Microcephaly

Subjects: Genetics & Heredity
Contributor: Catherine Yang

Autosomal recessive primary microcephaly (often shortened to MCPH, which stands for "microcephaly primary hereditary") is a condition in which infants are born with a very small head and a small brain. The term "microcephaly" comes from the Greek words for "small head."

Keywords: genetic conditions

1. Introduction

Infants with MCPH have an unusually small head circumference compared to other infants of the same sex and age. Head circumference is the distance around the widest part of the head, measured by placing a measuring tape above the eyebrows and ears and around the back of the head. Affected infants' brain volume is also smaller than usual, although they usually do not have any major abnormalities in the structure of the brain. The head and brain grow throughout childhood and adolescence, but they continue to be much smaller than normal.

MCPH causes intellectual disability, which is typically mild to moderate and does not become more severe with age. Most affected individuals have delayed speech and language skills. Motor skills, such as sitting, standing, and walking, may also be mildly delayed.

People with MCPH usually have few or no other features associated with the condition. Some have a narrow, sloping forehead; mild seizures; problems with attention or behavior; or short stature compared to others in their family. The condition typically does not affect any other major organ systems or cause other health problems.

2. Frequency

The prevalence of all forms of microcephaly that are present from birth (primary microcephaly) ranges from 1 in 30,000 to 1 in 250,000 newborns worldwide. About 200 families with MCPH have been reported in the medical literature. This condition is more common in several specific populations, such as in northern Pakistan, where it affects an estimated 1 in 10,000 newborns.

3. Causes

MCPH can result from mutations in at least seven genes. Mutations in the *ASPM* gene are the most common cause of the disorder, accounting for about half of all cases.

The genes associated with MCPH play important roles in early brain development, particularly in determining brain size. Studies suggest that the proteins produced from many of these genes help regulate cell division in the developing brain.

Mutations in any of the genes associated with MCPH impair early brain development. As a result, affected infants have fewer nerve cells (neurons) than normal and are born with an unusually small brain. The reduced brain size underlies the small head size, intellectual disability, and developmental delays seen in many affected individuals.

3.1. The gene associated with Autosomal recessive primary microcephaly

- ASPM

3.2. Additional Information from NCBI Gene

- CDK5RAP2
- CENPJ
- CEP152

- KNL1
- MCPH1
- STIL
- WDR62

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

- MCPH
- microcephaly primary hereditary
- primary autosomal recessive microcephaly
- true microcephaly

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