

Microcephaly

BIRTH DEFECT RESEARCH FOR CHILDREN



What is Microcephaly?

Microcephaly is a rare condition in which the circumference of an infant or child's head is smaller than normal. A child's head is considered abnormally small if it is two standard deviations from the median (average) size for their age and gender. In some cases, the forehead is narrow and sloping and the back part of the head is flat.



Microcephaly



Microcephaly may occur as an isolated defect or as part of a more complex syndrome. Diagnosis can be made through well-baby head measurements, X-rays, and ultrasound.

How many children are affected by Microcephaly?

Isolated Microcephaly that is not part of a more complex syndrome is very rare, about 1 in 250,000 births. The rate for Microcephaly that is part of a genetic, chromosomal or environmental syndrome depends on the incidence rate for those syndromes.

What causes Microcephaly?

Congenital Microcephaly is present at birth and is the result of abnormal brain development and growth during the first seven months of gestation.

Microcephaly may be caused by autosomal recessive inheritance or chromosomal abnormalities.

Autosomal recessive inheritance means that there is a genetic abnormality present in both parents.

Chromosomal abnormalities are adverse changes to the DNA in cells that can lead to birth defects.

Microcephaly may also be caused by a mother's infection during pregnancy with rubella (German measles), CMV (cytomegalovirus), or toxoplasmosis (parasite found in raw meat or cat feces). In addition, Microcephaly may be caused by environmental exposures during pregnancy such as alcohol, mercury and radiation.

Genetic Syndromes

The following syndromes may include Microcephaly:

- Aniridia-Wilms Tumor Syndrome
- Langer-Giedion Syndrome
- Johanson-Blizzard Syndrome
- Maternal PKU Fetal Effects
- Meckel-Gruber Syndrome
- Miller-Dieker Syndrome
- Rubinstein-Taybi Syndrome
- Seckel Syndrome
- Smith-Lemli-Opitz Syndrome
- Prader-Willi Syndrome

Environmental exposures

- Drug abuse by the mother
- CMV (Cytomegalovirus - fetal exposure to CMV herpes virus)
- Fetal Alcohol Effects (fetal exposure to alcohol)
- Fetal Aminopterin Effects (fetal exposure to the cancer drug, amniopterin)
- Fetal exposure to X-rays
- Fetal Methyl Mercury Effects (fetal exposure to methyl mercury through the mothers' consumption of fish containing mercury)
- Fetal Rubella Effects (fetal exposure to German measles)
- Toxoplasmosis (fetal exposure to a parasite contracted from undercooked meat or cat feces)



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Chromosome Abnormalities

These chromosomal conditions may include Microcephaly.

- Trisomy 13
- Trisomy 18
- Wolf-Hirschhorn Syndrome
- Cri du Chat Syndrome
- Partial deletion of long arm of 13

Helping a child with Microcephaly

The needs of a child with Microcephaly vary dramatically depending on the severity of the brain involvement and other associated conditions.

Medical team

Every child with Microcephaly should have a thorough evaluation to determine the severity of the condition and possible treatments. Doctors should also make every effort to find the specific cause. An evaluation may include blood tests, a CT scan, and amino acid screening. In some cases, a chromosomal evaluation may be done.

Special needs

Some children with Microcephaly may have few complications and minimal special needs. In other cases, special education and additional home care may be necessary.

Development

Children with less severe Microcephaly may have only minor intellectual impairment and nearly normal IQ's. Other children with more severe forms of impairment may be challenged with varying degrees of mental retardation, cerebral palsy, epilepsy, and visual handicap. Additional symptoms that are often associated with severe isolated Microcephaly include cataracts and short stature.

Psychological needs

Coping with the needs of a child with severe Microcephaly can be difficult for any family. A psychologist, mental health counselor, or social worker can help families work through their feelings and concerns, as well as help with finding additional resources for assistance.

Fact Sheet by:

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