CHILD syndrome

Description

CHILD syndrome is a condition that affects the development of several parts of the body. The name of the condition is an acronym of the major features: congenital hemidysplasia with ichthyosiform erythroderma and limb defects. The signs and symptoms of this disorder may vary from person to person, but they are typically limited to only one side of the body ("hemi-" means "half," and "dysplasia" refers to abnormal growth). The right side of the body is affected more often than the left side.

People with CHILD syndrome often have a skin condition characterized by large patches of skin that are red and inflamed (erythroderma) and covered with yellow, flaky scales (ichthyosis). This condition is most likely to occur in skin folds and creases and usually does not affect the face. The skin abnormalities are typically present at birth or appear within the first few weeks of life and may improve with time.

CHILD syndrome may also disrupt the formation of the arms and legs during early development. Some children with this disorder have shortened bones in the fingers or toes, while others have shortened or missing limbs. The limb abnormalities typically occur on the same side of the body as the skin abnormalities.

Some children have a curvature of the spine (scoliosis) or joint deformities that restrict movement (contractures). In some cases, CHILD syndrome affects the development of the brain, heart, lungs, and kidneys.

Frequency

CHILD syndrome is a rare disorder; fewer than 100 people with this condition have been reported in the medical literature. This condition occurs almost exclusively in females.

Causes

Variants (also called mutations) in the *NSDHL* gene cause CHILD syndrome. This gene provides instructions for making an enzyme that is involved in the production of cholesterol. Cholesterol is a type of fat that is produced in the body and obtained from foods that come from animals, particularly egg yolks, meat, and dairy products. Although high cholesterol levels are a well-known risk factor for heart disease, the body needs some cholesterol to develop and function normally both before and after birth.

Cholesterol is an important component of cell membranes and the protective substance that covers nerve cells (myelin). Additionally, cholesterol plays a role in the production of certain hormones and digestive acids.

The variants that cause CHILD syndrome change either the amount or the activity of the NSDHL enzyme, which disrupts the normal production of cholesterol within cells. Without enough of this enzyme, potentially toxic byproducts of cholesterol synthesis can build up in the body's tissues. Researchers suspect that low cholesterol levels can contribute to problems with the growth and development of many parts of the body. It is not known, however, exactly how a disturbance in cholesterol production leads to the specific features of CHILD syndrome.

Learn more about the gene associated with CHILD syndrome

- NSDHL

Inheritance

This condition is inherited in an X-linked dominant pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In females (who have two copies of the X chromosome), a variant in one of the two copies of the gene in each cell is sufficient to cause the disorder, although the signs and symptoms may be mild.

Researchers believe that affected males (who have only one X chromosome) have more severe signs and symptoms and typically die before birth.

Other Names for This Condition

- CHILD nevus
- Congenital hemidysplasia with ichthyosiform erythroderma and limb defects
- Congenital hemidysplasia with ichthyosiform nevus and limbs defects
- Ichthyosiform erythroderma, unilateral, with ipsilateral malformations, especially absence deformity of limbs

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Child syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265267/)

Genetic and Rare Diseases Information Center

- CHILD syndrome (https://rarediseases.info.nih.gov/diseases/6039/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Congenital hemidysplasia with ichthyosiform erythroderma and limb defects%22)

Catalog of Genes and Diseases from OMIM

- CONGENITAL HEMIDYSPLASIA WITH ICHTHYOSIFORM ERYTHRODERMA AND LIMB DEFECTS (https://omim.org/entry/308050)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28congenital+hemidysplasia+AND+ichthyosiform+erythroderma%5BTIAB%5D%29+OR+%28child+syndrome+AND+NSDHL%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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Last updated July 18, 2024