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CHILD SYNDROME: A RARE GENETIC DISORDER

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ABSTRACT

Congenital hemi dysplasia with ichthyosiform erythroderma and limb defects also known as CHILD syndrome is a genetic disorder with onset at birth seen almost exclusively in females. CHILD syndrome is a hereditary disorder transmitted as an X-linked dominant trait. The symptoms would appear at birth or shortly after birth. The combination of physical symptoms on the child would suggest they have CHILD syndrome. There is currently no treatment for CHILD syndrome so any treatment would target the symptoms currently present on Skin. Patients with left-sided involvement generally have more severe internal abnormalities, especially in regard to cardiac anomalies, and therefore have a worse prognosis. Early death in persons with CHILD syndrome is most commonly due to cardiovascular malformations. However, central nervous system, skeletal, kidney, lung, and other visceral defects can contribute to significant morbidity.

INTRODUCTION

CHILD syndrome is a rare Genetic disorder all most found in girls. The earliest recorded case of CHILD syndrome was in 1903. Otto Sachs was accredited for first describing the clinical characteristics of the syndrome in an 8-year-old girl. The nearest proceeding news on the topic was a report in 1948 by Zellweger and Uelinger, who reported a patient with "half-sided osteochondrodermatitis and nevus ichthyosiformis. The first case of CHILD syndrome with ocular manifestations in a patient suffering from progressive bilateral optic nerve atrophy was recently reported in 2010. CHILD syndrome is not fatal unless there are problems with the internal organs. The most common causes of early death in people with the syndrome are cardiovascular malformations. However, central nervous system, skeletal, kidney, lung, and other visceral defects also contribute significantly [1-4].

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Definition

Congenital hemi dysplasia with ichthyosiform erythroderma and limb defects (also known as "CHILD syndrome") is a genetic disorder with onset at birth seen almost exclusively in females. The disorder is related to CPDX2, and also has skin and skeletal abnormalities, distinguished by a sharp midline demarcation of the ichthyosis with minimal linear or segmental contralateral involvement [5].
Incidence:

Frequency

CHILD syndrome is a rare disorder with only 60 recorded cases worldwide till date. CHILD syndrome occurs almost exclusively in females.

Gender

Only 2 known cases have been reported in males, one having a normal 46,XY karyotype, suggesting an early postzygotic somatic mutation [6].

Age

Because CHILD syndrome is a congenital disorder, the

symptoms may be present at birth or may develop during the first few weeks of life and continue for the lifetime of the patient.

Causes

CHILD syndrome is a hereditary disorder transmitted as an X-linked dominant trait resultind from the faulty gene is on the long arm of the X chromosome (Xq28). Sterol-4-alpha-carboxylate 3-dehydrogenase, decarboxylating is an enzyme that is encoded by the *NSDHL (*dependent steroid dehydrogenase-like) gene This enzyme is localized in the endoplasmic reticulum and is involved in cholesterol biosynthesis. Mutations in the NSDHL gene are associated with CHILD syndrome which is a X-linked dominant disorder of lipid metabolism with disturbed cholesterol biosynthesis [7].

Pathophysiology

CHILD syndrome is inherited in an X-linked dominant fashion and is associated with a mutation of the NSDHL gene. This gene encodes for the enzyme 3beta-hydroxy sterol dehydrogenase which catalyzes a step in the cholesterol biosynthetic pathway. Locations of this enzyme include the membranes of the endoplasmic reticulum and on the surface of intracellular lipid storage droplets. A shortage of the enzyme may allow potentially toxic byproducts of cholesterol production to accumulate in the body's tissues. Mutations of the gene have been reported in all three types: missense, nonsense, and stop mutations, all resulting in loss of function of NSDHL. The type of mutation is not believed to be the underlining reason for clinical variations in the extent of involvement but rather the differences in the pattern of X inactivation. Although researchers suspect that low levels of cholesterol and/or an accumulation of other substances are responsible for disrupting the growth and development of many body parts, the precise rationale for the laterality of the syndrome has yet to be determined [8].

Sign and Symptoms

The acronym CHILD stands for the symptoms of the syndrome:

- CH-Congenital Hemidysplasia: One side of the body, most of the time the right side, is poorly developed. The right ribs, neck, vertebrae, etc. may be underdeveloped and the internal organs may be affected.
- I-Ichthyosiform Erythroderma: At birth or shortly after birth, there are red, inflamed patches (erythroderma), and flaky scales (ichthyosis) on the side of the body that is affected. Hair loss on the same side may also be possible.

LD -limb defects: Fingers on the hand or toes on the foot of the affected side may be missing. An arm or leg may also be shortened or even missing [9].

Table 1. Signs and Symptoms

Signs and Symptoms	Approximate number of patients (when available)	Percentage
Congenital ichthyosiform erythroderma	Obligate	100%
Aplasia/hypoplasia of the extremities	Very frequent	80%-99%
Epiphyseal stippling	Very frequent	80%-99%
Abnormality of the nail	Frequent	30%-79%
Hyperkeratosis	Frequent	30%-79%
Parakeratosis	Frequent	30%-79%
Abnormality of cardiovascular system morphology	Occasional	5%-29%
Adrenal hypoplasia	Occasional	5%-29%
Alopecia	Occasional	5%-29%
Aplasia/Hypoplasia involving the central nervous system	Occasional	5%-29%
Congenital hip dislocation	Occasional	5%-29%
Elevated 8(9)-cholestenol	Occasional	5%-29%
Elevated 8-dehydrocholesterol	Occasional	5%-29%
Flexion contracture	Occasional	5%-29%
Hearing impairment	Occasional	5%-29%
Hypoplastic pelvis	Occasional	5%-29%
Hypoplastic scapulae	Occasional	5%-29%
Micrognathia	Occasional	5%-29%
Pulmonary hypoplasia	Occasional	5%-29%
Renal hypoplasia/aplasia	Occasional	5%-29%

Scoliosis	Occasional	5%-29%
Short clavicles	Occasional	5%-29%
Short ribs	Occasional	5%-29%
Thyroid hypoplasia	Occasional	5%-29%
Vertebral hypoplasia	Occasional	5%-29%
Stillbirth	Very rare	1%-4%

Diagnosis

- The symptoms would appear at birth or shortly after birth. The combination of physical symptoms on the child would suggest they have CHILD syndrome.
- A skin sample examined under a microscope would suggest the characteristics of the syndrome.
- X-Ray of the trunk, arms, and legs would help to detect underdeveloped bones.
- CT scan would help detect problems of the internal organs [10].

Treatment

There is currently no treatment for CHILD syndrome so any treatment would target the symptoms currently present.on Skin.

- Dermatologic symptoms of CHILD syndrome are treated by applying skin softening (emollient) ointments, preferably plain petroleum jelly. This can be especially effective after bathing while the skin is still moist. Salicylic acid gel is another particularly effective ointment. The skin should be covered at night with an airtight, waterproof dressing when this ointment is used. Lactate lotion can also be an effective treatment for the skin symptoms of this disorder.
- Drugs derived from Vitamin A such as tretinoin, motretinide, and etretinate can be effective against dermatologic symptoms of CHILD syndrome. But can cause toxic effects on the bones in some cases. A synthetic derivative of Vitamin A, isotretinoin (accutane), when taken by pregnant women, can cause severe birth defects to the fetus. These Vitamin A compounds have not yet been approved by the Food and Drug Administration (FDA) for treatment of Ichthyosis.
- Other treatment is symptomatic and supportive.
- Genetic counseling may be helpful to families of patients with CHILD syndrome [11].

Possible Nursing diagnosis

- Impaired tissue perfusion r/t decrease cardiac output
- Impaired metabolism r/t hypoplastic endocrines
- Impaired skin integrity r/t scaly and red skin patches
- Body image disturbance r/t skin patches and short stature

Complications

CHILD syndrome is not fatal unless there are problems with the internal organs. The most common causes of early death in people with the syndrome are cardiovascular malformations. However, central nervous system, skeletal, kidney, lung, and other visceral defects also contribute significantly.

Prognosis

Patients with left-sided involvement generally have more severe internal abnormalities, especially in regard to cardiac anomalies, and therefore have a worse prognosis. Early death in persons with CHILD syndrome is most commonly due to cardiovascular malformations. However, central nervous system, skeletal, kidney, lung, and other visceral defects can contribute to significant morbidity.

STATEMENT OF HUMAN AND ANIMAL RIGHTS

All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

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Nil

CONFLICT OF INTEREST

None

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