Vereniging voor Ichthyosis Netwerken

Chanarin Dorfman Syndrome

Chanarin Dorfman Syndrome is a very rare skin condition. The fatty acids build up in the body, because they cannot be broken down. These fatty acids build up in the skin, but also in other parts of the body like the ears, eyes, muscles, liver and other organs. The Ichthyosis is caused by the accumulation of fatty acids in the skin. In 2009, 42 people around the world got diagnosed with this syndrome.

Chanarin Dorfman is an autosomal recessive genetic disorder. When both parents are carrier they have a 25% chance at each pregnancy of a child with the disorder. Parents carrying the faulty gene do not suffer from any of the symptoms and have normal skin. The syndrome is related to an abnormality in the ABHD5-gene. Because of the abnormality the body does not produce the substance that breaks down fatty acids. The fats, called triglyceride, build up in different parts of the body.

Because of the accumulation of fatty acids, the epidermisation process is disrupted. A red 'fish scale'-like skin appears like in other forms of ichthyosis. Sometimes the child is born with collodion membrane. Once shed, thickened skin will mostly appear in skin creases like knee and elbow.

The condition can affect the whole body and some people might have enlarged liver, clouding of the eye lenses, loss of hearing, poor coordination, weak muscles and intellectual disability. Because of the variety of problems a team of medical specialists should be involved in the treatment.

The severity will vary from case to case. Sometimes only the skin is infected. Sometimes multiple symptoms occur.

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