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My46 Trait Profile

Cutis marmorata telangiectatica congenita

Other Names: Van Lohuizen syndrome

Cutis marmorata telangiectatica congenita is a rare genetic skin disorder characterized by the presence of lace-like red or purple birthmarks on the body and sometimes other associated problems.

Characteristics of Cutis marmorata telangiectatica congenita

Cutis marmorata telangiectatica congenita (CMTC) is a rare genetic condition where the most common feature is the appearance of "cutis marmorata" – a deep red or purple birthmark in a mottled or fishnet-like pattern on the skin. Most of the time the birthmark is on the legs and/or trunk, but it can appear anywhere on the body. Some affected individuals also have another type of birthmark called capillary malformations, including port-wine stains or nevus flammeus (sometimes called a "stork bite"). Other skin changes include atrophy (thinning) or ulcerations. People with CMTC may have a difference in size between the two sides of their body – for instance, one leg smaller than the other. Some also have "webbing" of the fingers or toes (called syndactyly). A minority of individuals with CMTC develop glaucoma – an eye condition where there is increased pressure within the eye and progressive vision loss. Those individuals with glaucoma usually have cutis marmorata over the eye area.

The features of CMTC can be very different from person to person. There is no way to predict what problems a person with CMTC will have or how serious those problems will be. CMTC is rare and the number of people with CMTC is unknown; several hundred people have been reported in the medical literature. However, many people with CMTC are probably never diagnosed, so it may be more common.

Diagnosis/Testing

CMTC can be difficult to diagnose. There is no official list of signs or symptoms that must be present to say someone has CMTC. However, there is a list of suggested diagnostic criteria that is often used to help decide if someone has CMTC. If a person has all 3 major criteria (congenital marmorata, absence of venectasia, and unresponsiveness to local warming) AND 2 of the minor criteria (fading of marmorata within 2 years, telangiectasia within the marmorata area, port wine stain outside the affected area, ulceration within the affected area, or hypertrophy within the affected area) a diagnosis of CMTC is likely. The genetic etiology remains unknown.

Management/Surveillance

Management of CMTC is based on the signs and symptoms present. Affected individuals should have a full skin examination at least once a year. Evaluation by an ophthalmologist (eye doctor) may be recommended, especially if the face or eye is involved in the marmorata. Other specialists may include a dermatologist, plastic surgeon, or orthopedist. Treatment is not typically needed since the birthmarks usually fade over time, often during the first 3-5 years of life.

Mode of inheritance

CMTC is thought to be caused by a brand new (de novo) mutation in a yet unidentified gene.

Risk to family members

Because CMTC is a sporadic disorder that is not typically inherited, the risk of other siblings being affected is

thought to be very low.

Special considerations

None

Resources

CMTC-OVM
http://cmtc.nl/index.php?lang=en
Cutis marmorata telangiectatica congenital (CMTC)
http://www.cincinnatichildrens.org/health/c/ctmc/
Cutis marmorata telangiectatica congenital (CMTC) in children
http://www.childrenshospital.org/health-topics/conditions/c/cutis-marmorata-telangiectatica-congeita-cmtc

References

Kienast, AK. et al. (2009).“Cutis marmorata telangiectatica congenita: a prospective study of 27 cases and review of the literature with proposal of diagnostic criteria.” Clinical and Experimental Dermatology 34(3): 319-323.
Levy, R. et al. (2011).Cutis marmorata telangiectatica congenita: a mimicker of a common disorder.” Canadian Medical Association Journal 183(4): E249-E251.

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