Your Name

Instructor Name

Course Number

Date

Hypomelanosis of lto Syndrome

**Introduction**

Hypomelanosis of Ito is a genetic disorder. It is also called incontinentia pigmenti achromians and is a neurocutaneous disease (Jayaprasad, 2013). A patient suffering from it manifests skeletal, muscular and nervous system symptoms, and has hypopigmented macules on his/her body. Soles, palms, and scalps are those parts of the body which remain unaffected. The macules are usually on both sides of the body, however, the patches are not symmetrical. Patients with Hypomelanosis suffer from mental retardation, eczema, pes cavas, inverted cilia (of the eye), ventral hernia, back pain, reduced hearing, and much more.

It is not possible to arrive at any conclusions about extracutaneous symptoms, but there are some symptoms which cannot be ignored such as hypotonia, cramps, retardation and delayed development (Thomsen, 2014). Neurological problems associated with hypomelanosis of lto are found in 76% of patients of hypomelanosis of lto during the first ten years. Moreover, about 50% suffer from seizures or epilepsy.

In order to diagnose hypomelanosis of lto, evaluation with Wood’s lamp is most often used. Some additional genetic testing is also recommended. At present, there is no cure for the disease. Therapies are conducted which only treat the symptoms such as scoliosis and seizures. Moreover, usually, a multidisciplinary team provide the healthcare services to such patients. The prognosis depends on the intensity of symptoms. Patients having only skin symptoms usually live longer.

Hypomelanosis is more common in women than in men. The ratio is about 2.5:1. Moreover, about 1 in 10,000 people suffer from this syndrome. The symptoms usually appear in the first two years of their life. In medical literature, it was first mentioned in 1952.

**What genetic mutation is it made up of?**

Hypomelanosis of Ito is usually sporadic. In this context, sporadic means a new mutation as a result of which a genetic disorder has occurred for the first time in a family. It may also mean the appearance of a non-genetic disorder which is unlikely to recur in coming generations of a family. Family history of hypomelanosis of Ito (type skin lesions) exists in less than 3% of the patients. However, these cases are transmitted as autosomal traits. These autosomal traits are dominant. Nearly 10% of all patients of this disease have family history epilepsy and/or seizures.

In many patients of hypomelanosis of lto, not all cells have the normal 46 chromosomes, resulting in many abnormalities and mutation in genes. It may also result in extra material on a chromosome. Having extra material on chromosomes is called trisomy. Another possibility is mommy which is a loss of a certain portion chromosome. Monosomy is also called chromosomal translocation. Translocation involves the breaking off of certain portions of chromosomes and then their rearrangement. Consequently, genetic material is shifted and altered in the daughter cells. Some chromosomal abnormalities associated with hypomelanosis include chromosome 9q33 and chromosome Xp21.

**What chromosome or gene is affected in the body?**

In most cases, the causes of this disease cannot be identified (Janniger, 2014).. However, some cases have shown chromosomal abnormality. Mosaicism is reflected by the skin pattern sometimes. Mosaicism is a condition in which some of the skin chromosomes are normal and some have a gene or chromosomal abnormality, which results in dark and light areas appearing on the skin. Some cases also show alterations of X-chromosomes. Studies show that this alteration of X-chromosome along with mosaicism causes the said differences in the skin. It has also been observed that the in less than 3% of the patients, family history of hypomelanosis exists. In hypomelanosis of lto, the pigmentary phenotype may be attributed to karyotype abnormalities which disrupt the function of pigmentary genes.

# **References**

Janniger, CK. "Pediatric Hypomelanosis of Ito." (2014).

Jayaprasad, Sushmitha. "Hypomelanosis of Ito: A rare cutaneous syndrome ." 2013.

Thomsen, Christine Rohr. "Hypomelanosis of Ito presenting with pediatric orthopedic issues: a case report." *Journal of Medical Case Reports* (2014).