

Harlequin Ichthyosis - An Autosomal Disorder in Infants

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Abstract

Harlequin ichthyosis is an autosomal recessive very rare genetic disorder mainly seen in infants. This disease is mainly caused due to mutation in the gene for the protein ABCA12. The infants suffering with this disease have cracks and diamond shaped scales on the skin. This can be diagnosed by light microscopy, ultra structural findings etc. There are certain methods to treat this disease but there is no prevention. This article mainly deals with the symptoms, causes, epidemiology, pathogenesis, diagnosis, treatment and prevention of the Harlequin ichthyosis.

Keywords: Autosomal; Recessive; ABCA12 protein

Introduction

Harlequin ichthyosis/Harlequin baby syndrome/HI/harlequin foetus type is a very rare genetic disorder which is auto recessive and results in thickening of skin i.e., at birth the child's body is covered with an armour separated with deep cracks and diamond shaped plates are formed. Due to this eyes, ears, penis are contracted and also bacteria and other contaminants may enter the body of the child and cause fatal infection. This is a group of non-syndromic disorders of keratinization associated with a mutation in gene for the protein ABCA12. This can be diagnosed by fetal skin biopsy and analyzing amniotic fluid cells obtained by amniocentesis. This disease can be recognized by ultra sound. Constant care should be taken to moisturize and protect the skin. The frequency of the particular disease is 1 in 300,000 birth [1].

This abnormality affects the shape of eyelids, nose, mouth, ears and difficulty in movement of arms and diseases and may also cause respiratory failure. This disease disrupts the barrier making the infants to control water loss, regulate the body temperature and fight against the infections. The infants with this disease if not treated properly may lead to death of the infant [2].

Symptoms

- Thick skin plates that crack and split.
- Distorted facial features.
- Tight skin around eyes and mouth.
- Restricted breathing.
- Hands and feet that are small, swollen and partially flexed.
- Deformed ears or ears fused to the head.
- High blood sodium levels [3].
- Skin becomes scaly.
- Skin becomes dry.
- Anatomical changes like small head and deformed nose are seen [4].
- Hyperkeratosis [5].
- Limitation of joint mobility.
- Cataract.
- Dehydration [5].
- Foot and hand poly dactyly [5].
- Malignant hyper thalamia.
- Self injurious behaviour.
- Sudden cardiac death.

Causes

The cause of this disease is due to mutation in the ABCA12 gene which encodes a transporter protein which transports fats across cell membranes. Parents doesn't have any signs or symptoms of this disease but carry a copy of the mutated gene in their cells [3].

Epidemiology

The mortality rate of this disease is very high. More than 100 cases have been reported internationally. Some babies have survived the new born period because of intensive care and retinoid therapy but they are still in risk of systemic infection based on the sex there is no risk of harlequin ichthyosis. There is no known racial predilection of harlequin ichthyosis [6].

Pathogenesis

- All the patients suffering with this disease absent or defective lamellar granules and intercellular lipid lamella is absent.
- These granules secrete lipids that help in the maintenance of skin barrier at the interface between the granular cell layer and cornified layer.
- Due to the lipid abnormality there is excessive water loss in transepidermal layer which results in severe retention hyper keratosis [7].
- Patients with this disorder is caused by the mutation of inheritance in autosomes where it is a autosomal recessive disorder.
- The super family of ABC genes encodes proteins which transports substrates across the cell membrane. ABCA12 is to encode trans membrane protein which helps in liquid transport.
- This ABCA12 is to transfer of lipids from the cytosol of corneocyte into lamellar granules.
- Lamellar granules originates from golgi apparatus of keratinocytes in the stratum corneum which are responsible for secretion of lipids for maintaining the skin barrier at the interface between granular cell layer and cornified layer.
- The lamella from the skin's hydrophobic sphingolipid seal.
- The pivotal role in this disease supported by *in vitro* data.
- A milder form of ichthyosis is lamellar ichthyosis type-2 involves in mutation in the ABCA12 gene the phenotypic difference between the types of disorders is on the basis of variations in genotypic variance other chromosomal abnormalities are also described [6].

Diagnosis

This is diagnosed physically by examining the scales on the skin. If there is a doubt physician may test for skin biopsy. For early diagnosis pregnant mother can undergo ultra sound.

Methods

Light microscopy findings

- Extraordinary compact ortho hyper keratosis.
- Keratin plugs in hair follicles and sweat ducts.
- Absent lamellar and abundant vesicles in both the stratum granulosum and stratum corneum.
- Abnormal lipid droplets and vacuoles in the cytoplasm of keratinized cells in the thick stratum corneum.

Ultrastructural findings

- Abnormal lipid droplets and vacuoles in the cytoplasm of keratinized cells in the thick stratum corneum.
- Normal lamellar granules are absent in the cytoplasm of granular layer keratinocytes.
- In the extracellular space between the cornified cell and the granular cells lamellar structures are absent.

Biochemical analysis of skin samples

Family history for evidence [8].

Treatment

Air way maintenance

As this is a very rare skin disease the circulation and respiration of the neonatal is important. Medications are given through intravenous access to the babies. It is necessary to have cannulation of the baby's umbilicus and placed in an humidified incubator. After the signs have been stabilized the baby is transferred to neonatal nursery room.

Protection of conjunctiva

In order to protect baby's conjunctiva ophthalmic lubricants are applied. The babies affected with this disease should bath twice a day and should use sodium chloride wet compresses. To soften the hard skin bland lubricants are applied on the skin. Topical keratolytic medications should not be prescribed as they are toxic.

Intravenous access for feeding

Intravenous fluids are prescribed as they have feeding problems like excess cutaneous water loss. Serum electrolyte level monitoring is also used but there is a risk of having hypernatremic dehydration. Sterile environment is maintained to avoid infections [4].

Prevention

As it is an autosomal recessive genetic disorder which is inherited from parents to new born babies due to the mutation ABCA12 gene there is only cure but no prevention to this disease. There are many tests to diagnose and treatment to cure the disease than preventing the disease.

Conclusion

This disease is mainly seen in new born babies which forms diamond shaped scales and deep cracks. This disease is caused due to mutation in gene for the protein ABCA12 which may cause anatomical changes and dehydration. There are many ways to diagnose like skin biopsy. There are different tests and treatments but no prevention.

References

1. Harlequin-Type Ichthyosis (2016) Wikipedia.
2. Reference, Genetics. Harlequin Ichthyosis (2016) Genetics Home Reference.
3. Harlequin Ichthyosis (2016) WebMD Boots.
4. Harlequin Ichthyosis - Pictures, Treatment, Causes, Symptoms (2012) Ehealthwall.com.
5. <https://rarediseases.info.nih.gov/>

6. <https://ghr.nlm.nih.gov/condition/harlequin-ichthyosis>
7. (2016) Harlequin Ichthyosis Pathogenesis.
8. (2016) Images For Diagnosis Methods For Harlequin Ichthyosis.