ejpmr, 2021, 8(1), 346-348

SJIF Impact Factor 6.222

EUROPEAN JOURNAL OF PHARMACEUTICAL AND MEDICAL RESEARCH

www.ejpmr.com

Review Article

ISSN 2394-3211

EJPMR

HARLEQUIN ICHTHYOSIS – A RARE GENETIC SKIN DISORDERS

Kalyani Dhamal*, Omkar Bhalekar, Pragati Padhake, Vrushali Shelar, Anuja Misal

LSDP College of Pharmacy, Mandavgan Pharata, Tal- Shirur, Dist-Pune, Maharashtra, India 412211.

***Corresponding Author: Kalyani Dhamal**

LSDP College of Pharmacy, Mandavgan Pharata, Tal- Shirur, Dist-Pune, Maharashtra, India 412211.

Article Received on 13/11/2020 **Article Revised on 03/12/2020** **Article Accepted on 24/12/2020**

ABSTRACT

Skin disorder are vary in symptoms and severity. They may be minor, temporary or permanent, painless or painfull, life threatening. Harlequin ichthyosis is rare genetic skin disorder. It caused due to mutation in ABCA12gene. Gene ABCA12 has role in preparing protein which responsible for making skin cells. They transport lipids to superficial layers of skin. When ABCA is muted, skin barrier is distrupted. A newborn infant skin covered with thick plates of skin that crack and split apart. This genetic disorder is inherited in an autosomal recessive pattern. The tightening of skin became difficulty in breathing and eating. Infants born with harlequin ichthyosis have flat nose, feet and hand are small and swollen, ear are missing, and also joint mobility. It can be diagnosed at birth based on baby physical apperance. This review is focused on making awareness about rare genetic skin disorder.

KEYWORDS: Harlequin Ichthyosis, ABCA12,Genetic disorder.

INTRODUCTION

Ichthyosis is derived from Greek word" ichthys "means fish. It appeared as fish scale like skin. The Harlequin Ichthyosis is rare genetic disorder with harlequin baby syndrome, harlequin fetus, Ichthyosis fetalis as synonyms. It affect male and female in equal number. Approximately in 5,00,000 people 1 is affected or 7 cases in every year in united state. Harlequin ichthyosis is severe form of keratinizing disorders. It occurred in lethal during neonatal period, there is thickening of keratin skin layer. Recessive genetic disorder is occurred when individual receive one normal and one abnormal gene, this person is carrier for that disease but don't show any symptoms. The risk for two carrier parents to both pass abnormal gene, an affected child with 25% of each pregnancy. The chance for child to receive normal gene is 25%.

First case in world

First case in US reported in diary of Reverend Oliver Hart in 1750. Harlequin Ichthyosis is believed to inherited in an autosomal recessive manner. "armor" like thick yellow plates of scales with deep red fissuring. A new born infants was died in one day after birth. Fetal have respiratory infections, dehydration complications.

First case of Harlequin Ichthyosis in India

India recorded first case of harlequin ichthyosis in Nagpur. After a pre term a baby girl was born to young farmer couple in lata mangeshkar hospital. The entire body of baby covered with thick skin. The baby has no ear, fish like mouth, an inverted eyelid, two holes for nose. The baby was born at eight month pregnancy. There parents carry the mutated gene that causes skin disorder.

Symptoms

The thick plates make distort of facial fractures and restricted breathing and eating. Lamellar Ichthyosis have similar symptoms as harlequin ichthyosis. In which brown, dark plates like scale are separated by deep crack. They causes redness of skin and thickened of skin on palm and decreased sweating with heat tolerance. The infants with harlequin Ichthyosis have high risk for low body temperature, dehydration and elevated sodium level in blood. The symptoms of HI are:

- Eyelid turning inside out.
- Ear fused to head
- Nursing difficulty
- Breathing problems
- Hypernatremia
- Dehydration
- Low body temperature.

Causes

Harlequin ichthyosis is mainly caused by mutation in ABCA12 gene located on chromosome 2q34.ABCA12 gene makes the protein which is responsible for normal development of skin cells. The transfer of fats or lipids to outermost cell of body carried out by Proteins.

Tables

ABCA subfamily with their functions, disorder.

Member	Sites of Organ	Functions	Associated disorder
ABCA12	Skin epidermal keratinocytes	Lamellar granule lipid transport	Harlequin ichthyosis, lamellar ichthyosis
ABCA3	Lung, alveolar type 2 cells	Lamellar granule lipid transport	Fatal surfactant deficiency
ABCA1	Blood vessels	Cholesterol , phospholipid transfer	Tangier disease, early atherosclerosis.
ABCA4	Eye, retinal rod cells	Retinoid transport	Age related macular dystrophy, cone rod dystrophy
ABCA5	Skeletal muscle	--	--

Diagnosis

Harlequin Ichthyosis can diagnosed at birth place on baby physical appearance. Prenatal testing possible by testing fetal DNA for mutations in ABCA gene. During second trimester and onward features of harlequin ichthyosis seen on ultrasound. It can be diagnosed before birth using amniocentesis or chorionic villus sampling. The genetic testing registry provide information about genetic test for this condition.

Treatments

A neonatal intensive care team is required for caring the newborn with harlequin ichthyosis.

- Tube feeding help in malnutrition and dehydration.
- Retinoid prevent scaly skin
- Topical antibiotics prevent infection
- Covering the skin with bandages prevent infection.
- Artifial airway to improve breathing.
- Using lubricating eye drops or protective device on eyes

Managements

There is no proper treatments for harlequin ichthyosis (HI). Hence it is more important to care our skin.

- Protect the skin from environmental bacteria, viruses, fungi.
- Maintain the skin moist, clean. dry skin break out and valuranble to infections.
- Apply moisture rich product to skin.

DISCUSSION

Harlequin Ichthyosis (HI) is rare transferred genetic skin disorder. There are more than 20 types of ichthyosis with difference in their symptoms. Generally HI are found in North American, Asian, Mongolian groups. Family history is important to determine mode of inheritance. HI is genetically and phenotypically heterogeneous disorder. Frequency of HI is occurred as 1 in 3,00,000.

ACKNOWLEDGMENT

I am greatly thankful to my all dear friends and my college staff. And also gland thanks to my all supportive team. Also thanks EJMPR for accepting my short knowledge on Harlequin Ichthyosis.

CONCLUSION

Harlequin ichthyosis is rare genetic recessive disorder. Most commonly occurred in newborn infants. There is less chances for surveillance of fetal. More care with intensive team required for there caring. The prognosis of HI found as death in one month is relatively common. Still date there is special therapy or treatments for HI not found. June 2016 Nagpur reported the first HI case in India. This review is focused on generally rare cases of genetically skin disorders.

REFERENCES

1. James, William; Berger, Timothy; Elston Dirk (2005). Andrews Diseases of Skin, Clinical Dermatology.10th edition-Saunders ISBN 0-7216-2921-0.
2. Rapini, Ronald P;Bolognia, Jean L, Jorizzo, Joseph L.(2007). Dermatology: 2 volume Set.St.Louis:Mosby, ISBN 978-1-4160-2999-1.
3. Vergotine RJ; De Lobos, MR; Montero Fayad, M." Harlequin ichthyosis :a case report ". Pediator Dent., 2013; 35(7): 497-9.
4. Brittany G.Craiglow, MD:ichthyosis in newborn. Semin perinatol., 2013 Feb; 37(1): 26-31.
5. Hovnanian A.Harlequin Ichthyosis unmasked: A defect of lipid transport, J Din Invest, 2005; 115: 1708- 1710.
6. Fischer J.Autosomal recessive congenital ichthyosis, J Invest Dermatol, 2009; 129: 1319-2.
7. Hashemzadeh A, Heydarian F, Harlequin Ichthyosis. Acta Medi Iran, 2009; 47: 81-2.
8. Akiyama M. Pathamechanisms of harlequin ichthyosis and ABCA transporters in human diseases, Arch Dermatol, 2006; 142: 914-8.
9. https://rarediseases.info.nin.gov/diseases/6harlequinichthyosis , accessed May 1, 2019.
10. Lawlor,F and Pheris ,S.Harlequin fetus successfully treated with etretinate .Brit J Derm., 1985; 112: 585-90.
11. http://www.ichthyosis.eu.
12. Uitto J. The gene family of ABC transporters novel mutations, new phenotypes. Trends Mol Med., 2005; 11: 341-43.