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Harlequin Ichthyosis

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ABSTRACT: A mutation in the ABCA12 gene causes the unusual congenital skin condition known as harlequin ichthyosis. It is a skin condition that covers nearly the entire body of a baby and carries a high risk of serious, maybe fatal infection. The incidence of harlequin ichthyosis is about one in every 5,000 individuals. Both sexes are equally susceptible to this illness. The survival rate of affected babies varies from 10 months to 25 years, with the majority not making it past the first week of life. This condition, which causes a thick, dry fish-scale pattern all over the body, has no known cure.

I. INTRODUCTION:

Harlequin ichthyosis is an uncommon congenital skin disorder caused by a mutation in the ABCA12 gene. It's a skin ailment that affects almost the whole baby body and has a significant, potentially lethal infection risk. The prevalence of harlequin ichthyosis is around 1 in 5,000 people. This disease is equally likely to affect both sexes. Most affected babies do not survive past the first week of life; the survival rate ranges from 10 months to 25 years. There is no known treatment for this illness, which results in a thick, dry fish-scale pattern all over the body.

Case study:

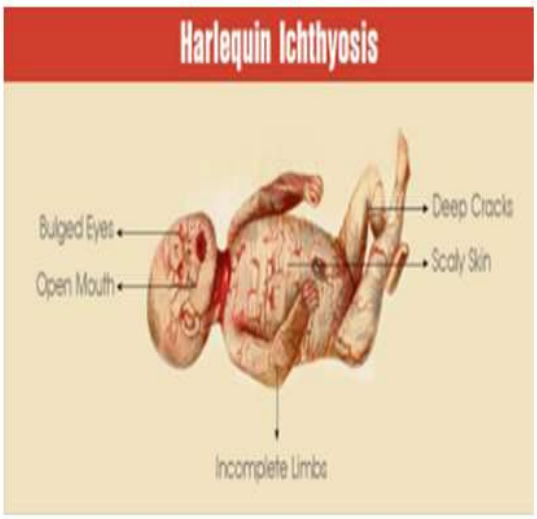
At 29 weeks and 4 days, a healthy spontaneous vaginal birth resulted in the birth of a female infant with Harlequin Ichthyosis weighing 1.6 kg. There was no family history of any inherited skin condition, and
the parents were estranged from one another. A physical examination showed that the skin was hardened, thicker, and divided unevenly by deep features in a yellowish, leathery white area.

Cause:

Harlequin ichthyosis is a genetic disorder that occurs due to a mutation in the ABCA12 gene. The gene plays an important role in the production of a protein that transports fats into the outer layer of the skin and helps in its development. Defect in the gene, therefore, does not allow the proper development of the skin, thereby explaining the appearance of the baby. If both parents have mutation of gene there is 25% chances that the child will be affected.

Symptoms:

1) Cracked and split thick skin plates
2) Misaligned facial features
3) Skin that is too tight around the eyes and mouth
4) Breathing difficulties (when the chest or abdomen are affected)
5) Small, swollen, and partially flexed hands and feet 6) Deformed or fused ears (ears may appear to be missing) 7) Elevated blood sodium levels



Diagnosis :

1) Physical examination: To determine the skin's normal appearance
2) Genetic testing: To identify faulty genes and validate the medical diagnosis
3) Sonography in the second and third trimesters
4) Amniocentesis: A genetic test that can be carried

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out early in pregnancy if a family history of a similar case has been documented

Complications :
1) External deformities: The child's ears are malformed and their skin is drawn tightly inward, giving them a distorted appearance. swollen eyelids that are difficult to open and shut.
2) Breathing: Tight, thick skin restricts a newborn's ability to move their chest, which causes respiratory issues.
3) Feeding: Due to limited suction and swallowing abilities, infants should be fed via a nasal tube in the early stages of their neonatal lives.
4) Dehydration: Affects how the body regulates its temperature and can lead to hypothermia
5) Infection: When pathogens penetrate the skin barrier during the early stages of life, life-threatening infections can result.

Treatment :
1) External deformities: The child has twisted ears and tightly drawn inward skin, which gives them an odd appearance. swollen, difficult to open and close eyelids.
2) Breathing: A newborn's inability to move their chest freely due to tight, thick skin results in respiratory problems.
3) Feeding: In the early stages of their neonatal lives, infants should be fed through a nasal tube due to their limited suction and swallowing abilities.
4) Dehydration: Impairs the body's ability to control its temperature and may result in hypothermia.
5) Infection: Life-threatening infections may arise from pathogens that breach the skin barrier in the early stages of life.

II. CONCLUSION:

From the Bove presentation it could be concluded that although there is presently no known treatment for this illness but supportive medical care may lengthen these patients survival . The mutation in the ABCA12 gene has been linked to Harlequin ichthyosis, genetic counseling and mutation screening of this gene should be considered, especially in families where there has been a consanguinity marriage.

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