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H SYNDROME:ARTHRITIS WITH POLYENDOCRIONOPATHY (AN UNUSUAL ASSOCIATION).

Rheumatology

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ABSTRACT

H syndrome is a rare autosomal recessive genodermatosis that falls under the histiocytosis-lymphadenopathy plus syndrome. The term "H syndrome" includes manifestations such as hyperpigmentation, hypertrichosis, hepatosplenomegaly, heart anomalies, hearing loss, hypogonadism, low height, and occasionally hyperglycemia.. The prognosis of H syndrome depends on several factors, including the extent and severity of clinical manifestations, the presence of complications, and timely diagnosis and management. Further studies are needed to explore the association between prognosis and the different mutations encountered in H syndrome. . In the background of Polyendocrinopathy with arthritis, we often tend to go for a genetic basis to pinpoint our diagnosis. Here in this review, we narrate a young girl with such illness who have been followed up for years and how a pediatric rheumatologist's liaisons with multiple departments for optimal care of the child diagnosed with 'H Syndrome'.

KEYWORDS

H syndrome, Histiocytosis, Arthritis

INTRODUCTION:

H syndrome is an autosomal recessively inherited form of histiocytos that occurs due to mutations in SCL29A3 gene that encodes human equilibrative nucleoside transporter 3 (hENT3).[1] The characteristic cutaneous findings seen in this syndrome are Hyperpigmentation, Hypertrichosis, and induration. The specific systemic manifestations associated are Hearing loss, Heart anomalies, Hepatomegaly, Hypogonadism, Hyperglycemia (diabetic mellitus), low Height (short stature), Hallux valgus (flexion contractures), and Hematological abnormalities.[2].. Here we present a cases of H syndrome presented to us in the age group of 5 year.

Case Report :

A 5-year-old girl presented with multiple joint pains involving the bilateral wrist, metacarpophalangeal joints (MCP), and PIP, knees, ankles, bilateral subtalar joints along with contractures of small joints of both hands and feet since a couple of years. She was born of a second-degree consanguineous marriage. History of death of 1st sibling at 8 years of age with unknown liver disorder and younger sibling currently aged 15 years diagnosed with type I diabetes mellitus since the age of 4 years. She was stunted for age. She was also noted to have syndromic facies. She also had a pes planus with calcaneo-valgus with crowding of toes and hyperextended great toe At the age of 13 years, she started having complaints of sticky stools with delayed onset of menarche (SMR stage III).

Investigations

Labs revealed anemia with ESR of 105 mm/1st h. She also had elevated levels of antithyroid peroxidase antibodies. Fasting sugar and lipid profile were normal. CT scan of the abdomen was suggestive of diffuse fatty infiltration of the pancreas. A further test for exocrine insufficiency could not be done due to financial constraints. Endocrine workup revealed low estradiol.

Management:

She was diagnosed to have polyarticular juvenile idiopathic arthritis with negative rheumatoid factor with autoimmune thyroiditis with short stature and delayed puberty. She was started on naproxen and methotrexate along with synthetic thyroid and estradiol supplement.

Genetic Study

A whole exome sequencing (WES) was sent across for both the sibling which revealed homozygous mutation in *SLC29A3* in exon3 c.400C>T [p. Arg134Cys] which was reported as being likely to be pathogenic causing H syndrome (cardiac anomaly, camptodactyly, clinodactyly, short stature, hepatosplenomegaly, hypergonadotropic hypogonadism , sensorineural hearing loss), PHID (pigmented hypertrichosis with insulin-dependent diabetes mellitus).

This case report shows the common clinical and investigation profile of H syndrome case admitted to IGICH hospital.

DISCUSSION

H syndrome is a novel form of histocytosis which involves multiple systems.the characterstic cutaneous features of the disorder are cutaneous hyperpigmentation overlying hypertrichosis and sclerodermatose thickening. Genetic mutuational analysis aids in conformation of the diagnosis when characterstic cutaneous changes are absent.mutations in SLC29A3 gene which encodes for the hENT3 resulting in this disorder besides the classical cutaneous features,a plethora of other systemic manifestations can be seen in H syndrome including low height,hearing loss,hyperglycemia,flexion contractures,hallux valgus,hepatosplenomegaly and various heart anamolies.among the various cardiac anamolies,pericardial involvement is seen to be the most common[3].

Hematological abnormalities including severe anemia with reticulocytopenia pancytopenia red cell aplasia and myelofibrosis.

Isulin dependent diabetes may be the sole presentation in few patient,pancreatic exocrine deficiency recurrent febrile episodes and complete agenesis of inferior venecava with varicose veins.proptosis or exopthalmous has been described as features.

Polyneuropathy, endocrinopathy, skin changes, arthritis were considered for the differential diagnosis.

CONCLUSION:

H syndrome is an extremely rarely reported entity more so in the Indian subcontinent with only 10 cases reported from india[4] .

We present this case to increase the awareness of this extremely rare and unique entity.

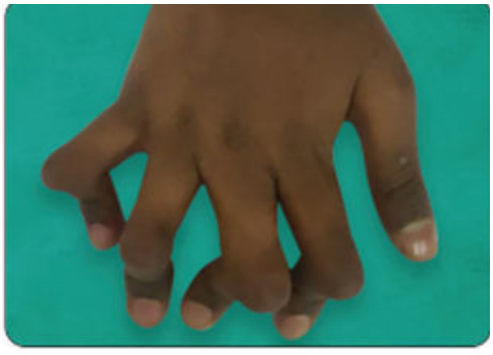

Fig 1: Camptodactyly of the fingers.

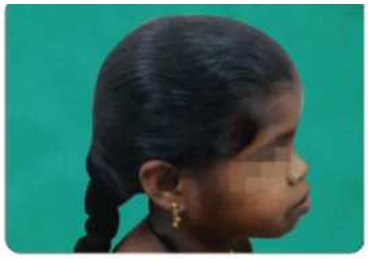

Fig 2: Depressed Nasal bridge/ Short neck/ Hypertelorism.

REFERENCES:

1. Molho-Pessach V, Lerer I, Abeliovich D, Agha Z, Libdeh AA, Broshtilova V, et al The H syndrome is caused by mutations in the nucleoside transporter hENT3 Amer J Human Genetics. 2008;83:529–34
2. Molho-Pessach V, Agha Z, Aamar S, Glaser B, Doviner V, Hiller N, et al The H syndrome: Agenodermatosischaracterized by indurated, hyperpigmented, and hypertrichotic skin with systemic manifestations J Am Acad Dermatol. 2008;59:79–85
3. Molho-Pessach V, Suarez J, Perrin C, Chiaverini C, Doviner V, Tristan-Clavijo E, et al The H syndrome: Two novel mutations affecting the same amino acid residue of hENT3 J Dermatological Science. 2010;57:59–61
4. Molho-Pessach V, Varma M, Godbole K, Kamath N, Zlotogorski A. H syndrome-Four new patients from India Indian J Dermatol Venereol Leprol. 2014;80:579