International Journal of Trend in Scientific Research and Development (IJTSRD)
Volume 7 Issue 1, January-February 2023 Available Online: www.ijtsrd.com e-ISSN: 2456 – 6470

Hunter Syndrome: A Case Report

Miss. Parimala L[1], Babu M[2]

[1]Professor cum Vice Principal, Saveetha College of Nursing,
[2]M.Sc Nursing (Nurse Practioner in Critical Care), Saveetha College of Nursing,
[1,2]Saveetha Institute of Medical and Technical Sciences, Saveetha University, Chennai, Tamil Nadu, India

ABSTRACT

Mucopolysaccharidosis type II (MPS II) or Hunter syndrome is Metabolism by lysosomal accumulation with a recessive inheritance pattern associated with the X chromosome. It is caused by lack of activity of the lysosomal enzyme iduronate-2-sulfatase, encoded by the IDS gene. Plasma iduronate-2-sulfatase enzymatic activity was measured and the IDS gene in genomic DNA was analyzed by automated direct sequencing. The enzyme activity was 1.2 _mol/l/h (reference value: >2 _mol/l/h) and molecular analysis detected the mutation c.1403G>A (p.R468Q), confirming the diagnosis of MPS II. rice field. In conclusion, there are few groups dedicated to this disease family here in Mexico, highlighting the need to form an expert team of physicians and scientists dedicated to inborn errors of metabolism to stay up to date.

KEYWORDS: *Mucopolysaccharidosis, Hunter syndrome, Turner syndrome, Iduronate 2-sulfatase, IDS gene*

How to cite this paper: Miss. Parimala L | Babu M "Hunter Syndrome: A Case Report" Published in International Journal of Trend in Scientific Research and Development (ijtsrd), ISSN: 2456-6470, Volume-7 | Issue-1, February 2023, pp.1096-1099, URL: www.ijtsrd.com/papers/ijtsrd52814.pdf

IJTSRD52814

INTRODUCTION

Mucopolysaccharidosis (MPS) is a group of autosomal recessive metabolic disorders caused by the absence or dysfunction of lysosomal enzymes required to degrade molecules called glycosaminoglycans (GAGs). These are long chains of sugar carbohydrates in all cells that help build bone, cartilage, tendons, calluses, skin, and connective tissue. Glycosaminoglycan's (formerly called mucopolysaccharides) are , which is also found in the fluid that lubricates the joints. Individuals with MPS do not produce enough or produce enzymes that do not function properly for any of the 11 enzymes necessary to break these sugar chains into proteins and simpler molecules. These GAGs accumulate in cells, blood, and connective tissue. The result is permanent and progressive cellular damage that affects physical appearance, physical performance, organ and system function, and, in most cases, mental development. Common clinical manifestations include facial dysmorphia, hepatosplenomegaly, joint stiffness and contractures, pulmonary dysfunction, myocardial hypertrophy and valvular dysfunction, and neurological involvement. Treatment has been mainly palliative as there is no effective treatment for MPS II (Hunter syndrome). However, enzyme replacement therapy (ERT) using recombinant human iduronate-2-sulfatase is currently being introduced. This case of MPS type II was identified because of its rarity and atypical features of mild mental retardation with normal intelligence, apical head, no corneal opacification, and all other features indicative of MPS type II. report. Therefore, not all MPS necessarily present with intellectual disability, corneal clouding, or atypical features such as: Head with polycephaly. The purpose of presenting this case is to highlight the characteristic symptoms of Hunter syndrome.

Case description:

This be alive a 5-year-old male patient, first pregnancy of healthy non-consanguineous parents, with a healthy 2-year-old sibling. Systemic hypertension was observed at his 3 months and psychomotor retardation was evident from his 16 months. Advice on patient behavior. A disorder characterized primarily by aggression. I was diagnosed with bilateral conductive hearing loss at the age of 3. He had a history of recurrent upper respiratory tract infections requiring hospitalization

on several occasions. He underwent a simple tonsillectomy at the age of four. A physical examination showed the patient to be frail, psychomotor-impaired, and in a severe general condition.

Malnutrition, orthocephaly, poor facial features, protruding frontal bones, sunken nasal bridge, wide nose with forward-slanted nostrils, thick lips, protruding ears, Short neck, low hairline, well-ventilated lung fields, regular heart sounds of good intensity, abdominal obesity with liver 2 cm below rib margin, umbilical hernia, male genitalia, extension of predominantly elbows and knees restricted limbs, clawed hands, and generalized hypertrichosis.

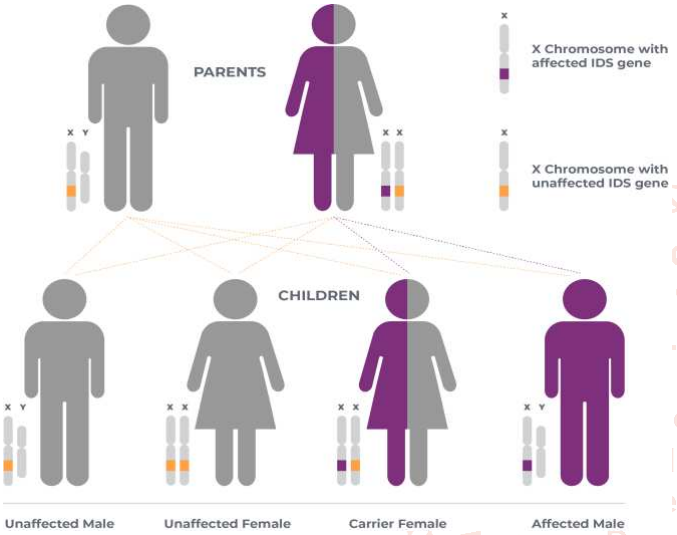

Figure 1: Hereditary causes of Family Tree

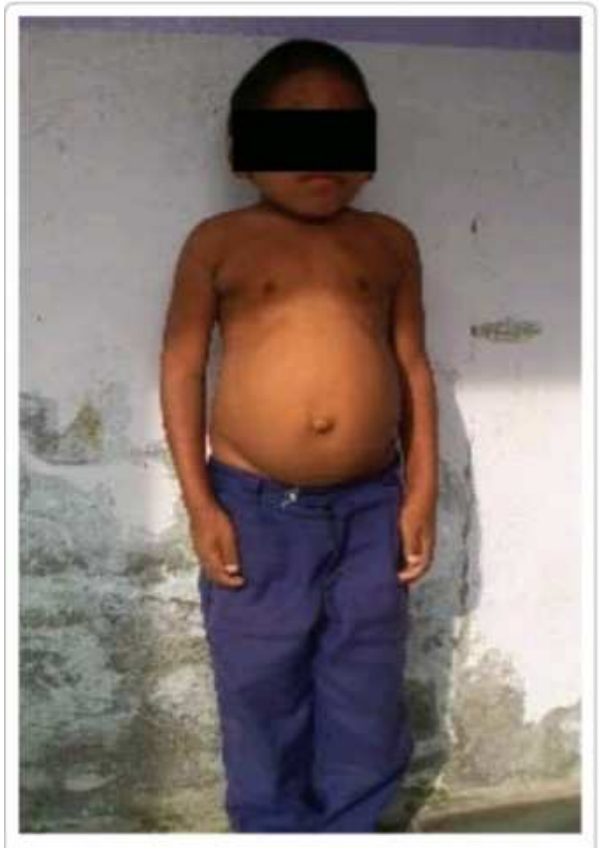

Figure 2: clinical picture of Hunter syndrome

Primary systemic manifestation and secondary systemic manifestations
hyperkinesis
Skeletal abnormalities (macrocephaly, enlarged chest, short stature)
Joint contractures
Hepatosplenomegaly
Cardiac valvular disease
Respiratory infections
Hirsutism
Abdominal hernias
Secondary systemic manifestations
Otitis media resulting in hearing insufficiencies and progressive deafness
Oral manifestations
Short and broad mandible
Macroglossia and protruding tongue
Peg lateral teeth
Anterior open bite
High arched palate
Hyperplastic gingiva
Generalised interdental spacing

Table1: Clinical Manifestation Hunter syndrome Investigation

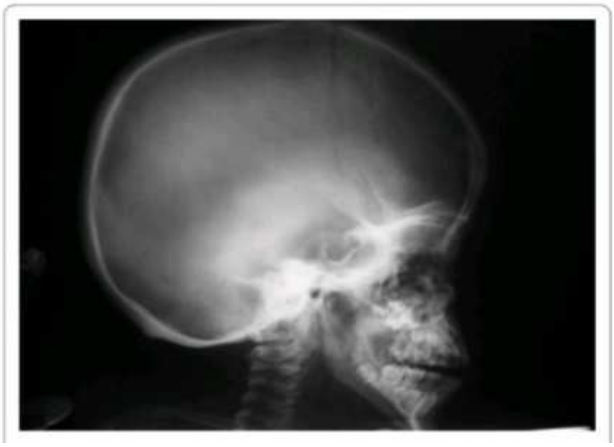

Figure 3: Anteroposterior and lateral x- ray of the skull showing `J 'Shaped sella turcica

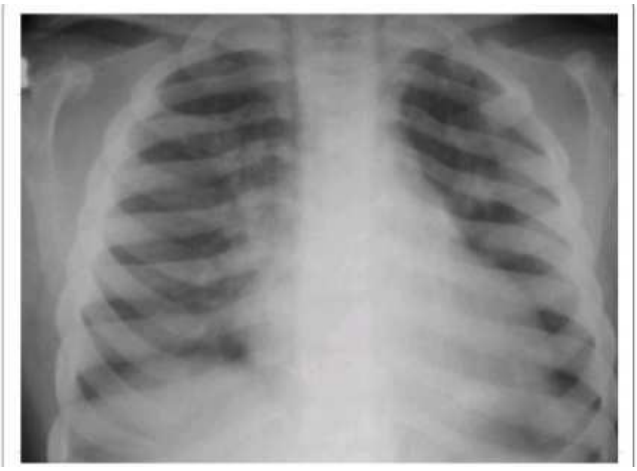

Figure 4: Anteroposterior chest x-ray showing paddle and spatulated ribs

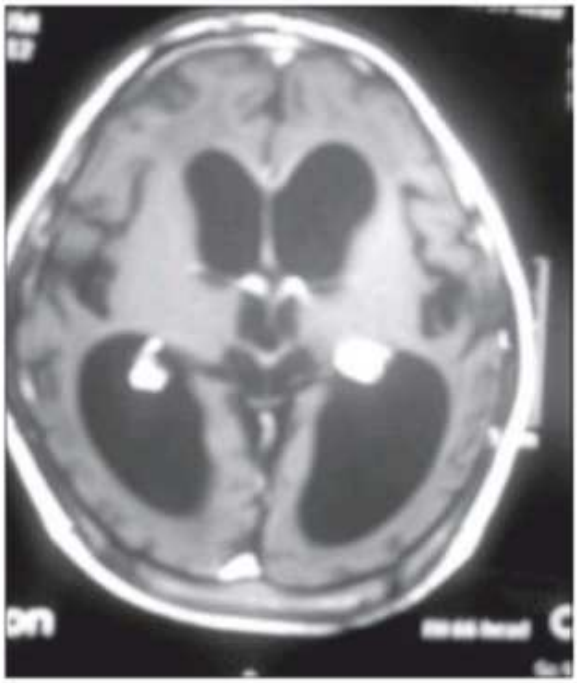

Figure 5: CT Brain showing communicating hydrocephalus

Chest X-ray and echocardiogram show no structural changes within normal parameters. EEG, abdominal ultrasonography, moderate hepatomegaly, normal bile, no epileptic activity. Genomic DNA was extracted from peripheral blood using a conventional method. The IDS gene was analyzed by polymerase chain reaction (PCR) and direct DNA sequencing. The resulting PCR products were sequenced on an ABI 3730 Automated Sequencer (PE Biosystems, Foster City, CA). Qualitative analysis of urinary glycosaminoglycans (GAGs) by positive toluidine blue. Plasma iduronatesulfatase enzyme activity decreased by 1.2 mol/l/h (reference value: >2 mol/l/h), confirming the diagnosis of MPS II. Molecular analysis of her IDS gene, which encodes the enzyme iduronatesulfatase, revealed that the patient was one such person. Mutation hemizygosity (c.1403G>A, p.R468Q). The patient died of respiratory complications 6 months after he was diagnosed with MPS II.

Treatment:

Enzyme replacement therapy (ERT)

Discussion:

Hunter syndrome is a rare genetic disorder and due to its progressive nature and the irreversible damage it causes, early diagnosis and therapeutic intervention are of paramount importance. Our patient could have been suspected of having this syndrome based on his clinical presentation, but no diagnosis was made during the course of his medical examination and admission. Recombinant human synthase (enzyme replacement therapy or ERT) is currently used to treat several lysosomal storage diseases, including Hunter syndrome. Major benefits of ERT include significantly reduced urinary GAG excretion with improved joint mobility, gait, pulmonary function, cardiac parameters and hearing, and reduced liver and spleen volumes. However, intravenous ERT does not cross the blood-brain barrier and cannot repair damage to the nervous system. Also, despite ERT, no ocular or skeletal problems or improvement in respiratory function have been reported, re-emphasizing the importance of early diagnosis. ERT has been shown to be safe in patients less than 1 year of age. Clinical evidence indicates that early intervention with ERT significantly improves patient quality of life and improves treatment response. The mutation found in the patient was first reported by Whitley et al. 1993; the guanine in exon 9 of the IDS gene is replaced by adenine, and her 468th glutamine in the protein is replaced by arginine. Sukegawa et al. Expression studies showed reduced enzymatic activity in fibroblasts from patients with this mutation. This mutation has been reported in multiple ethnic groups and is associated with a severe phenotype. Other mutations, R468W and R468L, have been reported in the same codon, suggesting that this site is a 'hotspot' for mutations in the IDS gene. To date, over 350 mutations in IDS genes have been reported. In conclusion, there are few groups dedicated to this disease family here in Mexico, highlighting the need to form an expert team of physicians and scientists dedicated to inborn errors of metabolism to stay up to date. there is. It is reasonable to assume that the vast amount of resources that social security systems and government programs allocate to these types of diseases will be used not only for medical care, but also for research and the generation of new medical knowledge.

Conclusion:

Mucopolysaccharidosis is a multisystem disorder which presents with a constellation of clinical findings. Careful and systemic approach is needed to accurately diagnose the exact type as enzymatic studies are not available in most centers.

Ethical disclosures:

Protection of human and animal subjects. The authors declare that no experiments were performed on humans or animals for this study.

Confidentiality of data:

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

Right to privacy and informed consent:

The authors declare that no patient data appear in this article.

Conflict of interest:
The authors declare that they have no conflict of interests.

References:

[1] Scarpa M, Almassy Z, Beck M, et al. Mucopolysaccharidosis type II: European recommendations for the diagnosis and multidisciplinary management of a rare disease. Orphanet J Rare Dis. 2011; 6:72.

[2] Kosuga M, Mashima R, Hirakiyama A, et al. Molecular diagnosis of 65 families with mucopolysaccharidosis type II (Hunter syndrome) characterized by 16 novel mutations in the IDS gene: genetic, pathological, and structural studies on iduronate-2- sulfatase. Mol Genet Metabol. 2016; 118:190---7.

[3] Muenzer J, Beck M, Eng CM, et al. Multidisciplinary management of Hunter syndrome. Pediatrics. 2009; 124:e1228---39.

[4] Muenzer J, Bodamer O, Burton B, et al. The role of enzyme replacement therapy in severe Hunter syndrome-an expertpanel consensus. Eur J Pediatr. 2012; 171:181---8.

[5] Guillen-Navarro E, Blasco AJ, Gutierrez-Solana LG, et al. Clinical practice guideline for the management of Huntersyndrome. Hunter Espana working group. Med Clin (Barc). 2013; 141:453.e1---13.

[6] Muenzer J. Overview of the mucopolysaccharidoses. Rheumatology (Oxford, England). 2011; 50 Suppl. 5:v4---12.