



Physiotherapy role in mucopolysaccharidoses-A case report

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Abstract

Mucopolysaccharidoses are group of metabolic disorders caused by absence or malfunctioning of lysosomal enzymes needed to break down molecules called glycosaminoglycan. A 8 years old boy had enlargement of the forehead, grey cornea and difficulty in moving the neck, raising both the arms, and with bended elbows, knees and ankles. Corneal clouding, coarse or rough facial features, Dwarfism and other skeletal irregularities with difficulty in walking and getting up from the floor from the 5 years of age.

MRI of brain showed macrocephaly, enlarged perivascular spaces of white matter, cervical spinal canal stenosis and NCV studies showed severe median nerve lesion on both sides leading to carpal tunnel syndrome. Tricuspid valve regurgitation & mild thickening of anterior leaflet of mitral valve was noted in echo-cardio graphy. The skeletal X-ray shows oval shaped vertebral bodies at L1, L2 and D12 minimal anterior luxation. He was diagnosed as Hurler's Syndrome (MPS- I) at one corporate hospital, Chennai.

At present the physical therapy measures like passive stretching for tightened hamstrings and tendoachilles, active exercises like strengthening of trunk and limb muscles were given to delay their joint stiffness problems and improve the ability to walk and running. Play therapy methods like throwing the air balls and cycling were also adopted Squatting as much as he possible, and stair climbing activities were encouraged to do daily. Within twenty days of physiotherapy the ROM in shoulder, elbow, wrist & knees were increased to complete his functional activities. His limitations have been improved. Follow up of 6 months were led to improve his squatting, walking & running capacity.

Individualized functional assessments charts, family counseling and importance of physiotherapy were given and encouraged to do all activities of daily living by some modifications to improve the quality of life. The multi-disciplinary team approach is lack in the rural or village community areas. These lag will also lead to poor improvement or follow up of the condition.

Keywords: mucopolysachardiosis, physiotherapy, hurler's syndrome

Introduction

Mucopolysaccharidoses are group of metabolic disorders caused by absence or malfunctioning of lysosomal enzymes needed to break down molecules called glycosaminoglycans (GAGs) [1]. These are found in the fluids that lubricate joints. The collection of GAGs in cells, blood and connective tissues will result in permanent progressive cellular damage which affects appearance, physical abilities, organ and system function and in most cases, mental development. It was estimated that 1 in 2,50,000 live born babies were presenting with this problem.

Types of Mucopolysaccharidoses

- MPS I
 1. Hurler syndrome
 2. Hurler-Scheie syndrome
 3. Scheie syndrome
- MPS II (Hunter syndrome)
- MPS III (Sanfilippo syndrome - A, B & C)
- MPS IVA & MPS IVB (Morquio syndrome)
- MPS V (Scheie syndrome)
- MPS VI (Maroteaux-Lamy syndrome)
- MPS VII (Sly syndrome)
- MPS IX (Natowicz syndrome)

Symptoms

- Coarse or rough facial features (flat nasal bridge, thick lips, and enlarged mouth and tongue)

- Dwarfism (abnormal bone size or shape) and other skeletal irregularities
- Corneal clouding
- Enlarged organs such as liver or spleen
- Excessive body hair growth
- Short and often claw-like hands, progressive joint stiffness with carpal tunnel syndrome
- Recurring respiratory infections are common with obstructive airway disease & obstructive sleep apnea
- Many individuals affected with enlarged or diseased heart valves¹

Diagnosis

Through Clinical examination and by urine glycosaminoglycans tests to find out excess mucopolysaccharides is excreted, amniocentesis and chorionic villus sampling can verify if a fetus is carrying a copy of defective gene. Regarding the long term outcome of mucopolysaccharidosis, the substance accumulations causing skeletal, respiratory, and cardiac manifestations are known to lead to poor prognosis, probably much more than other symptoms [2].

A 8 years old male brought to physiotherapy department by his parents with the presenting complaints of difficulty in raising both the arms, bended elbows, knees and ankles. He has the difficulty in walking and getting up from the floor for the past 1.5 years. He has been noted enlargement of the forehead, grey cornea and difficulty in moving the neck at

the age of 3 years and he was undergone for various investigations like Urinary Glycosaminoglycan test & Leukocyte enzyme analysis and he was diagnosed as Hurler's Syndrome (MPS- I) at one corporate hospital, Chennai.

The MRI brain done December 2012 showed macrocephaly, enlarged perivascular spaces of white matter & Corpus callosum, cervical spinal canal stenosis, and there was a severe median nerve lesion on both sides leading to carpal tunnel syndrome found by the NCV studies. Tricuspid valve regurgitation & mild thickening of anterior leaflet of mitral valve was noted in echo-cardiography. The skeletal X-ray shows oval shaped vertebral bodies at L1, L2 and D12 minimal anterior luxation.

Periodically he was on follow up with the hospital for further treatment and admitted for repeated respiratory tract infections. The MRI taken at January 2018 for brain & cervical spine showed non-obstructive hydrocephallus, Mild atlanto-axial sub-luxation with the kinking of cervico-medullary junction, indenting the cervical cord with bullet shaped vertebrae.

On examination child has coarse facies, muscle wasting noted in deltoid, arm, forearm, thigh, legs and foot muscles.

Short & stiff neck, ROM of both shoulder affected markedly with 40° of abduction and flexion, 30° terminal extension of both the elbows restricted and available movement in wrist was 30° in flexion & extension limited. Hand grip on both sides was fair and able to do the writing work given in the schools, but with deviated wrist.



Fig 1: Scapula Position while abduction



Fig 2: Protruded abdomen & Showing short neck



Fig 3: Radiological findings of wrist & fingers



Fig 4: Deformities of hip & knee noted on both sides. (Knock knee with flexion deformity)

Both hips and both knees were contracture in flexion deformity of 35°. He is able to walk & run with flexed hip & Knee for 10 to 12 meters. Bladder & Bowel, Balance & Co-ordination are normal. He finds difficult in single leg standing on the right side for few seconds compared with left side. And he also had limitation in full squatting to perform toilet activities, so advice were given to use the commode.

Treatment

Currently there is no cure for these disorders. Medical care is directed to treat symptomatic. Surgery to remove tonsils and adenoids are also done. Enzyme replacement therapies are also tried by BioMarin Pharmaceuticals, Shire Pharamaceticals Group to reduce the non-neurological symptoms and pain.¹ Bone marrow transplantation and Umbilical cord blood transplantation have had limited success in treating these conditions. Abnormal physical characteristics except for those affecting the skeleton and eyes, may be improved, but neurologic outcomes have varied and these therapies should be done after the extensive evaluation and counseling from the family members ^[3]. But, at present the physical therapy measures are needed to delay their joint problems and improve the ability to move. He has been on passive stretching of tightened hamstrings,

tendo Achilles. Passive and active exercises have been focused for both the upper & lower limbs, neck & trunk exercises. Squatting as much as he possible, cycling within his limitations and stair climbing activities were encouraged to do. Within twenty days of physiotherapy the ROM in

shoulder, elbow, wrist & knees were increased to complete his functional activities. His limitations have been improved. Follow up of 6 months were led to improve his squatting, walking & running capacity.



Fig 5: Cross leg sitting improved within 2 months of interval

Conclusion

The treatment of these types of conditions is focused on general and symptomatic, individualized functional assessments charts with appropriate treatment goals will rectify the child's problem for improving the quality of life. Moreover, the family members, relatives and society will also have their role to take care of the child like these rare diseases. Here the biopsychosocial model of disability approach has been implemented for these syndromes. Family counseling and importance of physiotherapy were given and encouraged to do all activities of daily living by some modifications.

The specific treatments currently available for mucopolysaccharidosis from enzyme replacement therapy, include hematopoietic stem cell transplantation, substrate reduction therapy, chaperon therapy, and gene therapy [2].

Even though the diagnosis & acceptance of this condition were being the difficult task for the parent, but the rehabilitation services or multi-disciplinary team approach is lack in the rural or village community areas. These lag will also lead to poor improvement or follow up of the condition.

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