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CASE REPORT

A Case of Mucopolysaccharidosis II /I-Cell Disease

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ABSTRACT

A 24-year-old second gravida was admitted at 17.3 weeks for MTP. She underwent a chorion villus sampling (CVS) at 13 weeks of gestation which revealed a fetus with normal karyotype but lysosomal enzyme assay revealed marked deficiency of beta-galactosidase consistent with I-cell disease. MTP was done. The fetus weighed 200 gm with no gross anomalies.

Keywords: Chorion villus sampling, I-cell disease, Beta-galactoridase.

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INTRODUCTION

Mucopolysaccharides which are otherwise called glucose amino glycans (GAG) are long chain sugary substances used in the building of bones, cartilages, skin tendons and many other tissues in the body. They form part of the structure of the body and also give the body some of the special features that make it work. For example, the slippery fluid that lubricate the joints contain mucopolysaccharides. In the course of daily life, there is continuous process of building new polysaccharides and breakdown of old ones. When this process is affected due to the deficiency of certain lysosomal enzymes which are responsible for the degradation of GAGs, they accumulate in tissues and hamper normal growth, development and functioning of the organs involved. There are various types of mucopolysaccharidosis (MPS) disorders depending on the enzymes involved namely: (a) Hurler syndrome-MPS type I, (b) Hurler Scheie syndrome—MPS type I H/S or I-cell disease, (c) Scheie syndrome–MPS type IS, (d) Hunter syndrome—MPS type II A and B, (e) Sanfilippo syndrome—MPS type III, (f) Morquio syndrome–MPS type IV A and B, (g) Maroteaux–Lamy syndrome—MPS type VI, (h) Sly syndrome—MPS type VII 1,2,3 subtypes.

CASE REPORT

A 24-year-old second gravida was admitted at 17.3 weeks for medical termination of pregnancy (MTP). She underwent a chorion villus sampling (CVS) at 13 weeks of gestation which revealed a fetus with normal karyotype but lysosomal enzyme assay revealed marked deficiency of beta-galactosidase = 20.56 µmol/g/hr (affected control 14.07. Normal controls 717.511) and alpha mannosidase = 8.55 µmol/g/hr (affected control 3.41, normal controls 154,65) consistent with I-cell disease.

CVS was advised as her previous child who was born at 38 weeks with intrauterine growth restriction, weighed 2 kg and normal apgar was diagnosed to be suffering from I-cell disease. The child had delayed milestones and was investigated and found to be hypothyroid and enzymatic analysis at 18 months age revealed I-cell disease.

MTP was done with 200 mg mifegest po followed by misoprostol 200 mg per vaginum.The fetus weighed 200 gm with no gross anomalies.

DISCUSSION

I-cell disease is an inherited lysosomal storage disorder.[1] It first was described in 1967 by Leroy and DeMars when they reported a patient with clinical and radiographic features similar to those of Hurler syndrome [mucopolysaccharidosis 1H (MPS 1H)] but with an earlier onset of symptoms and no evidence of mucopolysaccharidoria.[2] One unique feature of this disease was the presence of phase-dense intracytoplasmic inclusions in the fibroblasts of patients. These cells were termed inclusion cells, or I-cells; thus, the disease was designated I-cell disease. Spranger and Wiedermann subsequently classified this disease as mucolipidosis type II (ML II) because it had clinical characteristics that included mucopolysaccharidosis and sphingolipidoses.[3]

Etiology and Pathogenesis[1]

Abnormal transport of enzymes results in decreased levels of several enzymes in cells and excretion of enzyme into the extracellular compartment. N-acetylglucosamine-l-phospho-transferase is an essential enzyme for the synthesis of a mannose-6-phosphate recognition marker that targets lysosomal enzymes to the lysosome. It results in progressive accumulation of glycoprotein and glycolipids.

Clinical Description and Progression/Prognosis[1]

Progressive disorder with multiple organ and tissue involvement and wide spectrum of clinical severity. Presents in the 1st year of life. Rapid disease progression, with death occurring before 5 years of age. Clinical features include musculoskeletal skeletal deformities, coarse facial features, neurologic- hearing loss/deafness developmental delay rapid, progressive psychomotor deterioration, respiratory- recurrent and persistent upper respiratory tract infections, cardio-vascular—cardiac murmur to cardiac valvular and ischemic myocardial damage, ocular—mild corneal clouding, gastrointestinal severe gingival hypertrophy and hepato-splenomegaly.

Inheritance Pattern: Autosomal Recessive

Incidence: One in 325,000 live births (types II and III together).[2]
Diagnosis: Urine screening to estimate the levels of substances,

such as dermatan sulfate, heparin sulfate, keratin and chondroitin sulfate that are excreted, is the first step to identify the problem of MPS after clinical examination of the affected. Confirmation of the particular type of disorder is done by enzyme assays in white blood cells or serum of fibroblast culture (skin biopsy). Prenatal diagnosis of all types of MPS is possible by estimating the enzyme levels in chorionic villi biopsy done between 10 and 12 weeks and amniocentesis done between 16 and 20 weeks of gestation.

Conditions with similar presentations: GM1 gangliosidosis; mucolipidosis type I (Alpha neuraminidase deficiency-sialidosis); mucopolysaccharidosis type IH.[3]

Management: No disease-specific treatment available; one case study has explored intravenous pamidronate as an experimental option for bone pain.[4] Trials of Enzyme replacement therapy (ERT) are going on for MPS II and VI.

Other medical care: Symptom management.

Prognosis and Morbidity

Survival rate of the MPS affected individuals vary from early childhood to early adulthood and rarely some types like MPS Scheie has normal life span. Morbidity depends on the severity of the problem and it requires supportive medical care.

CONCLUSION

The incidence of MPS disorders is not known in Indian population. Considering the size of our population, many cases may go undiagnosed due to the difficulties in identifying the problems and confirmation of diagnosis. Help from support groups would indeed make a difference in the life of the affected and help them live their lives and not let the disease overshadow their existence.

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