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Two dimensional electrophoresis in diagnosis of lysosomal storage disorders mainly mucopolysaccharidoses in India

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Mucopolysaccharidoses are a group of inherited lysosomal storage disorders (LSDs) consisting of seven distinct clinical types and numerous subtypes. These are the result of deficiency of certain lysosomal degradative enzymes which are required to breakdown glycosaminoglycans (GAGs). The clinical features observed among MPS subtypes show overlapping signs and symptoms with other LSDs and rheumatologic disorders. This makes clinical diagnosis a challenge. India has witnessed an increase in the burden of lysosomal storage disorders in the recent years. Aim of the study was the temporal pattern of the disease from onset of symptoms to the final diagnosis. Retrospective analysis of the case record forms of the patients attending the hospital over a period of 5 years was under taken only the data of the patients who had confirmatory enzyme analysis or mutation study for LSD was further analyzed. The age at onset, suspicion of the illness, first clinical presentation to a tertiary hospital with genetics and the age at the final diagnosis of these confirmed cases were considered. A total of 480 patients were referred to the genetic clinic in this period. The diagnosis of LSD was suspected in 232 patients (9.08% of all referrals) and it could be confirmed in 119 cases (22.37% of the suspected cases). Maximum patients were diagnosed with Gaucher disease (31.93% of the diagnosed cases) followed by Mucopolysaccharidoses (20.16% of the diagnosed cases. Mutational analysis was available in 22 patients (18.48% of the diagnosed cases. With the advent of new therapies, appropriate medical management is possible and hence establishing timely diagnosis has become crucial. In the retrospective data analyses done in our institute two different diagnostic approaches were discussed. The diagnostic approach includes GAG quantification followed by Two Dimensional cellulose Acetate Electrophoresis and enzyme analysis which seems to be appropriate for diagnosis MPS I, II, IV, VI and VII. The second diagnostic approach involves direct enzyme analysis which is pertinent for MPSII as these patients have a distinct clinical phenotype which enables clinician to establish the diagnosis with ease. The relative frequency of MPS I, II, IIIA, IIIB, IIIC, IVA, IVB and VI observed in our experience is 14%, 8%, 19%, 16%, 4%, 27%, 3% and 9% respectively. If family history of LSD is known initially, the pregnant women with gestation period between 16-22 weeks can be tested for GAG quantification in Amniotic fluid. With these two method combined it will be a useful, fast and cost effective diagnostic tool.

Biography

Gujjarlapudi Deepika has completed her MD Biochemistry from Osmania Medical College and presently pursuing her PhD in Asian Institute of Gastroenterology, Hyderabad India.

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