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Screening for Mucopolysaccharidoses Among Mentally Retarded Children

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ABSTRACT 350 mentally retarded children from the Coastal districts of Andhra Pradesh were screened for various inborn errors of metabolism. Three children –2 cases of Hunter's syndrome and 1 case of Sanfilippo syndrome were found to have mucopolysaccharidoses (MPS).

INTRODUCTION

Muccopolysaccharidoes (MPS) are a groups of genetically inherited lysosomal storages of glycosaminoglycan (CAG) metaolism due to the absence of expression of a specific lysosomal enzymes, resulting in imperfectly catabolised CAG and their excessive storage in tissues and also excretion in urine. There are specific regions of the polymeric CAG chain. The gradual staorage of incompletely metabolised CAG causes marked distortion of many tissues with consequent severe somatic changes. The deposition of lipids and mucopolysaccharides (MPS) in brain cells leads to mental and neurological deterioration resulting in mental retardation Suzuki (1972). The inheritance in all forms of MPS is autosomal recessive (Suzuki, 1972). The inheritance in all forms of MPS is autosomal recessive except in Hunter's syndrome, which is X-linked. The present paper reports cases of mucopolysaccharidoses detected from 5 coastal districts of Andhra Pradesh.

MATERIAL AND METHODS

The subjects for the present study were children below 15 years drawn from schools and special institutions for mentally retarded children from different areas of Vizianagaram, Visakhapatnam, East Godavari, Krishna and Guntur districts of Andhra Pradesh, Sounth India. The identification of mentally retardation children was mainly based on IQ tests developed by American Association on Mental Deficiency (Ingalls, 1978).

Urine samples were collected and the following tests were done to confirm the cases. Toludine blue spot and CTAB turbidity tests were the priliminary chemical screening tests used and the final confirmation has been done using cellulose acetate electrophoresis to characterize the different mucopolysaccharides following Lewis et al. (1974).

RESULTS AND DISCUSSION

Out of 315 mentally retarded children screened, 3 (0.95%) were found to have the MPS. They include cases of Hunter's syndrome and 1 cases of Sanfilippo syndrome; the salient clinical and biochemical features of these cases are listed in table 1.

From India, a few reports are available relating to MPS (Abraham et al., 1969, 1970; Ganguly et al., 1986; Kagalwala, 1988a,b; Verma, 1986; Puri and Verma, 1978; Rao, 1991). As for Andhra Pradesh, Krishna et al., (1992) reported 12 cases out of 100 families (1.2%).

It is of utmost importance that the affected persons/families are detected at the earliest possible age in order to initiate suitable therapy/management and also to prevent the recurrence of the condition in the family, appropriate genetic counselling. Due to certain unusual social practices like consanguinity prevailing in most of the rural populations of South India there may be many more children affected with

Table 1 : Clinical and biochemical profiles with mental retardation

Patient	*Sex*	*Age*	*Clinical symptoms*	*Consanguinity*	*IQ*	*Biochemical profiles in urine*	*Disorder*
DP	Male	6	Coarse facies, thick eyebrows, short stature hepatosplenomegaly umbilical hernia, no corneal clouding, mental retardation	Consanguineous (MBD)	25	Heparan & Dermatan sulfate	Hunter's syndrome
UP	Male	5	Coarse facies, thick eyebrows, short stature hepatosplenomegaly, no corneal clouding, mental retardation	Consanguineous (FSD)	25	Heparan & Dermatan sulfate	Hunter's syndrome
LB	Female	6	Coarse facies, thick eyebrows, short stature hepatosplenomegaly umbilical hernia, no corneal clouding, mental retardation	Consanguineous (FSD)	30	Dermatan sulfate	Sanfilippo syndrome

these disorders who go undiagnosed.

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