Indian Journal of Psychiatry, April 1987, **29**(2), pp. 169-170

BRIEF COMMUNICATIONS

SOME PARADOXIAL OBSERVATIONS NOTED IN THE PREVALENCE OF HURLER'S AND HUNTER'S SYNDROME

H.S. NARAYANAN[1]
K.S. MOHAN[2]
N. JAYASIMHA[3]
B.S. SRIDHARA RAMA RAO[4]

Hurler's and Hunter's syndromes belong to the group of lyosimal storage disorders where the mucopolysaccharides (MPS) are the storage substances (Stanbury et al. 1983). Both Hurler's and Hunter's cases show the excretion of dermatan and heparan sulphates in the urine. Despite this similarity in biochemical feature the two differ in some other respects. For instance corneal opacity is noted only in subjects with Hurler's but not in Hunter's syndrome. Also the nature of inheritance is autosomal recessive in Hurler's while it is sex linked in Hunter's. During our studies on subjects with cases with MPS excretion we noted a variation in the prevalence of the two types and the present paper briefly reports the same.

Table
The age and sex distribution of the cases

Age Group	Hurler		Hunter		Total	
(in years)	M	F	M	F	M	F
0 – 3	14	6	1	–	15	6
4 – 6	8	3	7	–	15	3
7 – 9	6	2	2	–	8	2
10 – 14	–	1	4	–	4	1
Total	28	12	14	–	42	12

The subjects of this study were obtained from patients attending the Mental Retardation clinic at this centre. Based on clinical and biochemical investigations about eighty cases were found to belong to the group of mucopolysaccharidoses. Among them forty were Hurler's type and fourteen were Hunter's type.

From the clinical observations it was noted that cases of Hunter lived somewhat longer than Hurler. It might, therefore be expected that the prevalence of Hunter's should be more frequent than Hurler's.

In order to test this the Handy-Weinberg formula was used to find out the gene frequency. In the case of Hunter's the gene frequency was similar to the frequency of affected males.

Both Hunter and Hurler do not have a long span of life and so the question of reproducing does not arise. The mutation rate for both Hurler and Hunter syndromes is assumed to be the same since, equilibrium exists between addition of mutant genes and loss of mutant genes due to failure to reproduce. By using the Hardy-Weinberg formula under conditions of equilibrium, $q = \mu$, where q is the gene frequency and μ is the mutation rate for the Hurler gene.

1. Associate Professor of psychiatry
2. Resident in psychiatry
3. Research assistant in neurochemistry
4. Professor and head of the department of neurochemistry
National Institute of Mental Health and Neuro Sciences, Bangalore 560 029, India.

In case of Hunter q is the same as frequency of affected males and can exist on X chromosome of the female carrier who is asymptomatic.

Then the theoretical expectation that frequency of Hunter should be more than Hurler's, since they live longer is not borne out.

Our findings are similar to the ones reported by McKusick (1970). McKusick has explained the observations on the following lines i) A prejudice that autosomal recessive conditions are more prevalent than sex linked ones, ii) Diagnostic criteria, iii) Age at which cases are seen (Hurler's features are noted earlier than Hunter's), iv) Consanguinous marriages apparently carry an increased risk for prevalence of Hurler's.

The present observations would point out the need for early detection of cases so that some management measures could be planned (such as control of upper respiratory tract infection) and also to take suitable measures to offer genetic counselling. In some centres abroad, in high risk mothers prenatal detection by amniotic fluid examination and termination of pregnancy in indicated cases is advocated.

References

McKUSICK V. A. (1970), The relative frequency of the Hurler and Hunters Syndrome, *New England Journal of Medicine,* 283, 853-854.

STANBURY JB., WYNGAARDEN JB., FREDERICKSON DS., GOLDSTEIN JC & BROWN MS. (1983), The metabolic basis of inherited disease, 5th Edn., McGraw Hill Co., New York.