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Experience with Mucolipidosis Type IV in Newfoundland. C. Prasad[1], D. J. Buckley[2], C. Pushpanathan[3], E. J. Ives[1], E. Goldin[4] and R. Schiffmann.[5] [1]Division of Medical Genetics,[2]Pediatric Neurology Program,[3]Department of Pathology, Janeway Child Health Centre, St. John's, Newfoundland, Canada, [4,5]Developmental and Metabolic Neurology Branch, NINDS, NIH, Bethesda, MD.

Mucolipidosis type IV is a unique autosomal recessive progressive neurodegenerative disorder of unknown etiology. There is minimal facial dysmorphism and absence of organomegaly. Most patients reported are of Ashkenazi Jewish descent.

We present two patients from northern Newfoundland with significant developmental and speech delay, ataxia, corneal clouding, and absence of organomegaly. The skin biopsy was significant for the presence of lamellar inclusions which exhibited autofluorescence characteristic of Mucolipidosis IV cells. Cranial MRI showed changes in signal intensity consistent with increased ferritin deposition in the thalamus in one of the patients (female) but was normal in the second (male). Both the patients did not show any changes in the white matter, nor hypoplasia of the corpus callosum. Both patients had elevated serum gastrin levels, 936 and 332 pmol/L (0-43 pmol/L) respectively. The finding of high serum gastrin in patients with Mucolipidosis IV has been recently reported by Schiffmann R et al (Proc Natl Acad Sci USA 1998 Feb 3;95(3):1207-12). This finding in combination with the characteristic skin biopsy findings can be diagnostic for this disorder.

Our patients belong to the same geographical area. They are not of Ashkenazi Jewish ancestry. Both the patients exhibit a milder course without neurological deterioration. They lack the typical cranial MRI findings of cerebellar atrophy and hypoplastic corpus callosum reported recently.

With the availability of reliable diagnostic testing, the pathogenesis and etiology of this poorly understood condition will likely be delineated in the future.

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Study of Some Morphological Physiological and Serological Parameters in Twins Monica Singh*, S.P. Singh* Harsurinder Kaur* *Deptt. of Human Biology, Punjabi Univ. Patiala, Punjab.

The present study was conducted on 18 pairs of twins. The similarity method was used for zygosity determination. Eight pairs were found to be monozygotic and the rest dizygotic.

Intra-pair differences regarding various morphological and physiological parameters were observed in these twins. MZ twin pairs showed very small differences for morphological parameters than DZ, confirming that MZ twins have similar genetic makeup. Physiological parameters however showed comparatively larger intra pair differences as they depend more on emotional state of the individual. The mean maternal age was found to be 28 years in our sample. The twinning tendency was found to be inherited in 50 percent of the cases with paternal transmission being more common (38.8 percent) than maternal.

None of the mothers admitted the intake of fertility drugs. No heritable diseases were detected though in a few cases one of the pair was reported to be more prone to some allergic or infectious conditions.

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Heterogeneity of ApolipoproteinE polymorphism and its Isoforms in Five caste groups of Punjab, India, Puneetpal Singh*, P.P. Balgir**, I.J.S. Bansal**, *Deptt. of Human Biology, **Deptt. of Biotechnology, Punjabi University, Patiala, Punjab.

Genetic polymorphism of ApolipoproteinE is controlled by three common alleles(E2,E3 and E4) and some rare alleles (e.g. E1, E4v etc;) at Apo E structural gene locus and may be demonstrated by Iso-electric Focussing, Density gradient poly-acrylamide gel electrophoresis and Immunoblotting techniques et;. The common ApoE isoform E2 (arg158-cys) and E4 (cys112-arg) differ functionally from the parent E3 isoform, explaining their effects on the normal variation of Plasma Lipoprotein concentrations and their association with Hyperlipidemic conditions. Apolipoprotein genotypes were determined in five endogamous caste groups in a sample of 150 Indians from Punjab. ApoE have been observed at respective allele frequencies 0.077, 0.807 and 0.115 in these population groups of Banias, 0.050, 0.825 and 0.125 in Brahmins, 0.057, 0.846 and 0.097 for Jat Sikhs, 0.060, 0.800 and 0.140 in Khatris and 0.035, 0.839 and 0.125 for mixed group 'Others'. Apo E3 (ApoE3,3 and ApoE3,4) subjects had higher frequencies than ApoE4 (ApoE4,3 and ApoE4,4) and ApoE2 (ApoE2,2 and ApoE3,2) subjects in all the groups studied suggesting the low prevalence of Alzheimer's disease and CHD's. The estimated ApoE allele frequencies were combined with data from other studies of the World to outline the total map of ApoE allele frequency distribution from Europe to Asia.