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Ashtavakra Journal Club

Session 2022-2023

B.Ed. Spl. Edu. LD

Date: 25/11/22 **Time:** 2:00 PM

TOPIC- Genetics of Intellectual Disability

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NAME OF THE JOURNAL (APA)- H Ropers. (2008). Genetics of intellectual disability, Studies in Education, vol. 18, Issue 3, pages 241-250.

ABSTRACT

Objective: To study the genetics of intellectual disability from different IQ tests and getting a quantitative isolation of mutation.

Methods: By Standardized IQ test, Wechsler Adult Intelligence Scales (WAIS), Wechsler Intelligence scales for Children (WISC)

Results: With the help of standardized IQ tests, cytogenetically visible chromosomal aberrations account for almost 15% of all cases. Deletions and duplications that are too small to be detectable by conventional karyotyping seem to be equally important, previously overlooked causes of ID, as discussed below. X-linked gene defects are thought to be responsible for ~10% of the ID found in males, which means that there must be other factors to explain why cognitive impairment is far more common in males than females [11] (see also Skuse [12] and Nguyen and Disteche [13]). The cause of ID is still unknown in up to 60% of the cases [14•]. This leaves ample room for autosomal gene defects, either novel mutations giving rise to isolated cases with dominant forms of ID, or recessive forms, most of which will also appear as sporadic cases in the small families that are characteristic of industrialized countries.

Despite widespread prenatal diagnosis in older mothers, Down syndrome (trisomy 21) remains the most important single cause of ID. Severe dominant forms of ID are rarely familial because, in general, affected

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individuals will not reproduce. Little is known about the prevalence of dominant ID, but given the high proportion of apparently relevant *de novo* CNVs in cohorts of patients with 'idiopathic' ID, it cannot be rare. On the contrary, functional considerations as well as epidemiological data suggest that the majority of the gene defects that give rise to disease will be inherited as recessive traits.

Conclusion: Until recently, the function of most ID genes has remained largely obscure, but during the past two years, remarkable progress has been made in this field. Fragile X syndrome, the most common heritable form of ID that is caused by loss of function of the RNA-binding FMR1 protein FMRP, is thought to result from upregulation of group1 metabotropic glutamate receptor (mGluR) signalling [76]. Numerous recent studies have shed lighter on the role of FMRP at the synapse and in dendritic MRNA.

Using commercially available next-generation sequencing systems, re-sequencing of >30 megabases of genomic DNA has become possible in a single experiment (reviewed by Bentley [114]), and oligonucleotide arrays have been employed for the quantitative isolation of DNA from defined genomic intervals [115, 116]. Combination of these methods should greatly speed up the search for mutations in large deletion or linkage intervals.

Keywords: IQ test, chromosomal, parental diagnosis, down syndrome, fragile X syndrome.

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