Summary
To identify the genetic cause of intellectual disability, study subjects (n=130) were recruited and multiple assays were employed to screen the whole genome.

Severe intellectual disability was identified in 12% of the subjects and 11% were moderately disabled, based on IQ evaluation. A clinical evaluation of developmental delay was identified in 77% of subjects.

Conventional cytogenetic analysis by high resolution GTG banding is a whole genome approach and revealed that 3% of study subjects showed chromosomal abnormalities (three deletions and one inversion).

PCR-based screening of FMR1 gene, a characteristic feature of fragile X syndrome, did not detect any mutations in the study population.

Subtelomeric rearrangements examined with FISH were detected at a frequency of 7.7% and two balanced rearrangements were detected in the parental samples.

MLPA, used to evaluate interstitial chromosomal rearrangements, yielded a frequency of 2%. No microdeletions/microduplications were detected using the QMPSF technique.

Though targeted and specialized techniques permitted the detection and delineation of more rearrangements, phenotype and genotype correlation was not observed in majority of the study subjects.