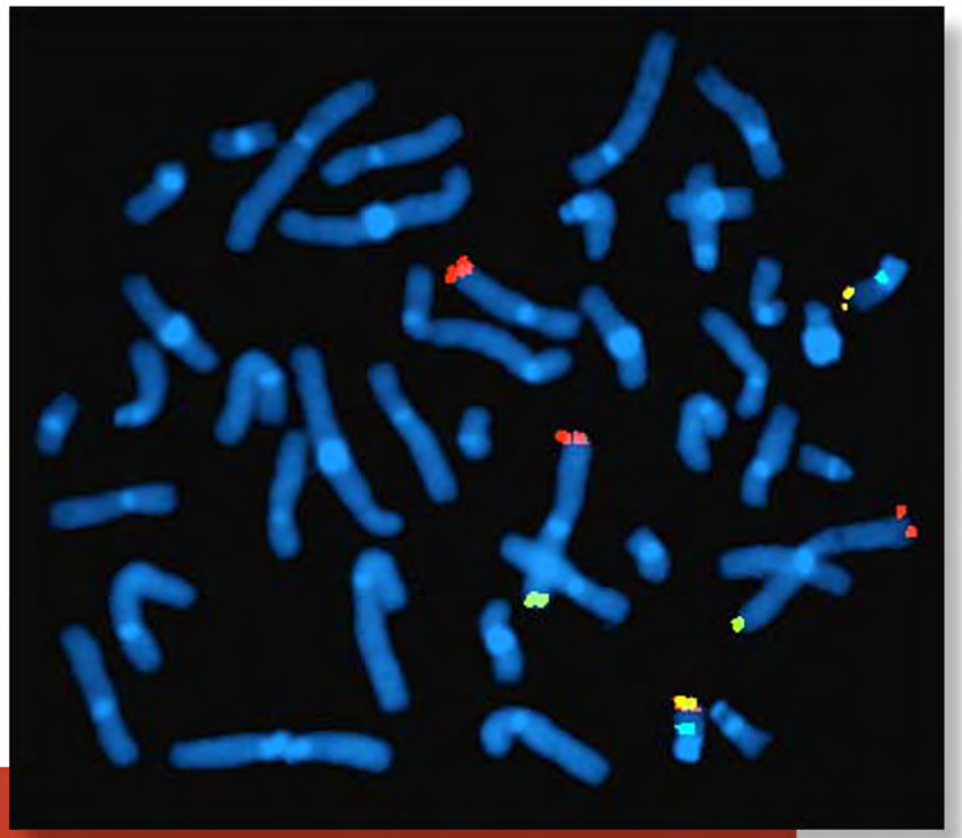

Summary

- 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.