

High Expression Levels and Amplification of rRNA Genes in a Mentally Retarded Child with 13p+: A Familial Case Study

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Abstract : A cytogenetic and molecular genetic study of the family with a male child who had mental retardation and autistic features revealed an abnormal chromosome 13 bearing an enlarged p-arm with amplified ribosomal DNA (rDNA) in a boy and his father. Cytogenetic analysis using standard G-banding and FISH with labeled rDNA probes revealed an abnormal chromosome 13 with an enlarged p-arms due to rDNA amplification in a male child, who had clinically confirmed mental retardation and an autistic behavior. This chromosome is evidently inherited from the father, who has morphologically the same chromosome, but is healthy. The karyotype of the mother was normal. Ag-NOR staining showed brightly stained large whole-p-arm nucleolus organizer regions (NORs) in a child and normal-sized NORs in his father with 13p+-NOR-amount mosaicism. qRT-PCR with specific primers showed highly increased levels of 18S, 28S and 5,8 S ribosomal RNA (rRNA) in the patient's blood samples compared to a normal healthy control donor. Both patient's father and mother had no elevated levels of rRNAs expression. Thus, in this case, rRNA level seems to correlate with mental retardation in familial individuals with 13p+. Our findings of rRNA overexpression in a patient with mental retardation and his parents may show a possible link between the karyotype (p-arm enlargement due to rDNA amplification), rDNA functionality (rRNA overexpression), functional changes in the brain and mental retardation. The study is supported by Russian Science Foundation Grant 15-15-10001.

Keywords : mental retardation, ribosomal DNA-rDNA, ribosomal RNA-rRNA, nucleolus organizer region-NOR, chromosome 13

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