In a male patient with developmental delay, autistic behaviour, obesity, lymphedema, hypertension, macrocephaly, and facial features of chromosome 5p duplication (trisomy 5p) a 3.7 Mb de novo tandem microduplication of 5p13.1-13.2 (rs4703415-rs261752, i.e., chr5:35.62-39.36 Mb) was identified. This observation contributes to the characterization and dissection of the 5p13 duplication syndrome. The possible role of increased NIPBL gene dosage is discussed.
Occurences:
- Einrichtungen > Fakultäten > Fakultät für Medizin > Kliniken und Institute > Institut für Humangenetik > 2011

entries: