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In this study we present data on the temporal bone computed tomography of rare case of inner ear malformation - Michel aplasia in patient with congenital bilateral sensorineural deafness from Russia (Sakha Republic, Yakutia, Eastern Siberia). The CT-images (1 mm) of temporal bone has been performed. This case of Michel aplasia was characterized by symmetrical agenesis of the cochlea and semicircular canals, bilateral abnormality of the facial nerve canal, and abnormality of the internal auditory canal on both sides. Results of this study confirm high diagnostic importance of the temporal bone computed tomography for detailed characterization of developmental abnormalities of inner ear.

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J11.27

Interstitial del(3)(p26.3p26.1) in a patient with deletion 3p syndrome

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Partial deletion of chromosome 3p is a rare disorder with variable chromosomal breakpoints and consequently phenotypes. Although most of these deletions involve the 3p terminus, interstitial deletions may also give rise to features of the syndrome.

We report the case of male patient with mental retardation and minor dysmorphic features and a 7.4 Mb deletion of 3p26.3p26.1 [arr 3p26.3p26.1 (61,891 7,527,372) x1], encompassing the genes CHL1, CNTN6, CNTN4, CNTN4-AS2, IL5RA, TRNT1, CRBN, LRRN1, SETMAR, SUMF1, ITPR1, EGOT, LOC100507582, BHLHE40, ARL8B, EDEM1, MIR4790 AND GRM7. We compare the clinical phenotype of this patient to previously reported cases of 3p syndrome.

Microarray technologies are increasingly becoming the tool of choice to accurately determine the underlying genetic cause and resulting phenotype in patients with mental retardation and multiple anomalies. aCGH allows to precisely determine the length and breakpoints in order to better understand a child's future development and needs.

In the present case molecular karyotyping has characterized a 3p deleted region with haploinsufficiency of neurodevelopmental genes associated with cognitive deficit and mental retardation, which may help to identify genes important to growth and development that contribute to the deletion 3p syndrome phenotype and aid in better understanding the molecular basis of the 3q syndrome.

J11.28

16p13.11 microduplication: a case report

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The short arm of chromosome 16 is very rich in segmental duplications, predisposing this region of the genome to a number of recurrent rearrangements, namely deletions and duplications. Although it is already known that there is a strong association between 16p13.11 deletion and neuropsychiatric disorders, the clinical significance of its reciprocal duplication is not clearly defined yet. 16p13.11 microduplication that results of non-allelic homologous recombination is a very rare genetic alteration which can be associated with variable clinical features including behavioural abnormalities, developmental delay, congenital heart defects and skeletal anomalies.

We report a 7-years-old boy with global developmental delay, speech absence, microcephaly, dysmorphic facial features and inexpressive facies. Microarray analysis revealed a 3.3Mb duplication comprising the 16p13.11-p12.3 region, which was confirmed by fluorescence *in situ* hybridization with a BAC clone for 16p13.11. Eight annotated genes are present in this region including *NDE1*, the candidate gene for neurological and behavioural phenotype. Although this microduplication has been found in the normal population, is significantly enriched in patients with autism, schizophrenia and cognitive impairment. Several case reports until now suggest that this genomic abnormality has incomplete penetrance and variable expressivity and can constitute a new syndrome. With this case we intend to contribute to expand the spectrum of clinical findings associated to this genomic abnormality and provide further knowledge of the pathogenic involvement of this duplication.

J11.29

Immune deficiency in monosomy 18p

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Immune deficiency may be a rare feature in chromosomal disorders. A 31-year-old man with immunodeficiency, intellectual disability and facial anomalies was admitted to the genetics department. The patient was born to nonconsanguineous parents. He had congenital hypothyroidism and intellectual disability. During childhood, he had recurrent oral aphthous lesions, ear infections and pneumonia. Investigations revealed IgA, IgG and IgM deficiencies with a normal lymphocyte count. Magnetic resonance imaging of brain showed hyperintense lesions in peripheral white matter. On physical examination, he had short stature, and dysmorphic facial characteristics including flat nasal bridge, low-set ears, epicanthic folds, and short philtrum. Facial dysmorphic features, intellectual disability and immunodeficiency may suggest some dysmorphic syndromes including 22q11.2 deletion, ICF and others. Karyotype analysis revealed a partial chromosome 18p deletion: 46,XY,del(18)(p11.1).

Chromosome 18p partial deletion is one of the most common deletion syndromes. The estimated frequency is 1 in 50,000 live-born infants. The cause of this disorder is deletion of short arm of chromosome 18 or sometimes deficiency in a ring 18 chromosome. IgA deficiency, central nervous system abnormalities such as holoprosencephaly, and mild to severe intellectual disability usually accompany.

J11.30

A report of partial monosomy of distal 5p and partial trisomy of distal 19q in a family with Charcot-Marie-Tooth disease

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We present two sisters with mild developmental delay/ intellectual disability and additional clinical features. The younger girl, 21 months of age, had hypotonia, short stature, microcephaly, hip dysplasia, delayed teeth eruption, bilateral epicanthus, wide nasal bridge, short philtrum, short neck, clinodactyly of fifth finger, low serum calcium and parathormone. Clinical features in her sister, 11 years of age, were similar, but in contrast, she had macrocephaly, and additional skeletal anomalies: kyphosis, pectus carinatum, contractures of elbows and knees, right clubfoot. Positive family history of Charcot-Marie-Tooth disease was revealed (patient's mother had duplication of CMT1A region, patient's grandfather had clinical symptoms of CMT). Testing for PMP22 duplication was performed, and it was positive for the older sister. Karyotype of both sibs established by GTG banding was 46,XX, der(5)t(5;19)(p15.3;q13.2). Subsequent chromosome analysis in parents revealed maternal balanced translocation t(5;19)(p15.3;q13.2), which was later confirmed by FISH. The same translocation was detected in grandmother.

Both del5p and dup19q are described in literature as pathogenic imbalances. 5p terminal deletion causes cri-du-chat syndrome (mewing cry, microcephaly, epicanthus, depressed nasal bridge, fifth finger clinodactyly, hypotonia). 19q13.2qter duplication is associated with wide range of congenital anomalies and dysmorphic facial features, including growth retardation, microcephaly, heart defects, hypoplasia of the gallbladder, renal anomalies, hypertelorism/flat nasal bridge, dysplastic ears, downturned mouth corners, clinodactyly. Synthesis of del5p15.3 and dup19q13.2 symptoms determines the clinical phenotype of our patients. Additional skeletal malformations in older sister are caused by CMT. This report provides clinical characterization of previously unreported chromosomal rearrangement.