significant implications for clinical genetic counseling, screening and management strategies for affected families.

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A NOVEL MUTATION IN PERICENTRIN (PCNT) GENE CAUSED MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM

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Microcephalic primordial dwarfisms (MOPDs) are autosomal recessive disorders characterized by prenatal and postnatal growth retardation, microcephaly and distinct facial features with prominent beaked nose, receding forehead, and micrognathia. The most common type is MOPD type II and it's characteristic features are specific growth pattern with very severe IUGR, adult height below 110 cm, neonatal proportioned head size later progressing to microcephaly, absent or mild mental retardation, typical bone dysplasia and small loose secondary dentition. Here we report a 6 months old boy with MOPD II syndrome diagnosed by clinical and radiographic findings and confirmed by mutation analysis. As far as we know ten genes have been identified for microcephalic primordial dwarfism, encoding proteins involved in fundamental cellular processes including genome replication, DNA damage response, mRNA splicing and centrosome function. We find a novel premature stop codon mutation in the PCNT gene. Hence, the diagnosis of MOPD by mutation analysis can be a valuable method for patients. Furthermore, in the near future, other genes could be identified in patients with MOPD.

SOS1 ASSOCIATED NOONAN SYNDROME PRESENTING WITH SUPERNUMARY TEETH; A CASE REPORT

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Noonan syndrome (NS) is a common autosomal dominant disorder which is characterized by chest deformation, congenital heart disease, short stature and distinctive facial features. Severe gingivitis and supernumerary teeth are rarely seen in connection with NS. In this report, a 12-year-old boy with Noonan syndrome whose oral examination revealed an anterior open bite, supernumerary teeth, severe anterior gingival enlargement, dental crowding associated with diesterna, tapered incisors, narrow high-arched palate and prominent rugae was presented.

DNA diagnostics of MIDD and MELAS syndromes in Slovakia

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Background. The two syndromes, MIDD (Maternally Inherited Diabetes and Deafness) and MELAS (Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis and Stroke-like episodes) arise on a common genetic cause – a mutation in mtDNA, most often the m.3243A>G transition in the gene *MT-TL1* for $tRNA^{Leu(UUR)}$. This mutation leads to different clinical symptoms according to the heteroplasmy levels in different tissues. Usually, the first presentation of the MIDD syndrome is a progressive bilateral sensorineural hearing loss emerging in adolescence, meanwhile, diabetes develops mostly between 30 – 40 years of life. The MELAS syndrome has a more severe progression with further neurological and metabolic symptoms.

Patients and methods. The aim of our study was to search for the m.3243A>G mutation among patients fulfilling at least one of the following criteria: maternal inheritance, conjoint diabetes and hearing impairment, diabetes development after 25th year of life, and progressive hearing loss. DNA of 255 unrelated probands was extracted from peripheral blood and/or buccal mucosa and analyzed for presence of m.3243A>G variant using RFLP and/or Real-Time PCR. DNA testing was also extended to the family members of probands carrying the mutation.

Results. The m.3243A>G mutation was found in 8 probands and 10 of their family members. Probands' phenotypes varied from diabetes as the sole symptom to a complex picture of the MELAS syndrome (in one proband). All of the probands (100%), but only in 3 of 10 relatives with the mutation (30%), developed diabetes or impaired glucose tolerance (with onset age from 21 to 54 years). Five probands (62.5%), and 4 (40%) of the relatives with the mutation had hearing impairment (with onset age from 16 to 59 years). Three family members, aged 18, 28 and 33 years (buccal heteroplasmy levels 58, 33 and 15%, respectively), were

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