polyneuropathy associated with bilateral striatal necrosis. Using homozygosity mapping the pathogenic homozygous missense mutation c.373G>A was identified. This mutation alters a highly conserved glycine residue at position 125 to serine in the *SLC25A19* gene which encodes the mitochondrial thiamine pyrophosphate transporter. *SLC25A19* mutation was previously reported only in the Amish congenital lethal microcephaly but our patients' phenotype is markedly different with normal head circumference, early childhood development, and age-appropriate cognitive skills. Based on the role of *SLC25A19* in mitochondrial thiamine transport, supplementation with high dose thiamine was initiated in all patients. Preliminary results suggest a favorable effect.

Our report suggests an allelic phenotype in *SLC25A19* gene defects. Determination of the *SLC25A19* sequence may be warranted in patients with bilateral striatal necrosis or progressive polyneuropathy.

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A NOVEL DE NOVO MUTATION OF CHROMOSOME 7 [46,XX,DEL(7)(P14.2 P15.1)] IN A CHILD WITH FEEDING PROBLEMS

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Background: The phenotype and severity of symptoms associated with mutations of chromosome 7 are directly proportional to the number of bases away. Distal and interstitial deletions are described in 40 cases.

Aim: To report a child with a novel de novo mutation of chromosome 7 [46,XX,del(7)(p14.2 p15.1)].

Clinical case: Female, born at 38 weeks with RCIU and feeding problems with episodes of cyanosis after feedings and failure to thrive. Physical examination showed low implantation of ears, hypertelorism,

oblique palpebral fissures, retrognathia, and palate ogved, insertion anomalies of the toes, poor facial expression and mild axial hypotonia.

Atransfontanelar ultrasound, MRI, bronchofibroscopy and metabolic study were normal. She was hospitalized until the 32th day of life.

She starts speech-language therapy and presented improvements in swallowing. The PEG was removed at 36 months.

She had recurrent UTI with normal DMSA but with a vesicoureteral reflux (grade III). She has taken UTI antibiotic prophylaxis.

Imagiological studies reveal a bilateral osteonecrosis of femoral epiphysis (Legg-Calvé-Perthes disease).

Nowadays (4years-old) she is being normally fed (BMI=15.8kg/m²). She has a mild delay of psychomotor development impairment (GQ: 74.1) and some speech problems.

Conclusion: This is the first case report of a patient with this de novo mutation of chromosome 7. This rare mutation was a cause of severe feeding problems in the first years of life.

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THE INFLUENCE OF DOWN SYNDROME ON DEVELOPMENT OF PRIMARY (DECIDUOUS) TEETH

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Down Syndrome (DS) (Trisomy 21) is a relatively common anomaly occurring in one in every 600-700 live births and characterized by impaired growth and development and early senescence. Signs of abnormal development have been observed on ultrasound as early as the first trimester. By the second trimester additional signs are evident on ultrasound, affecting skeletal growth as well as other organ systems. Dental anomalies include reduced tooth size and number, thin enamel and abnormal crown form as well as taurodont roots and a high frequency of missing teeth. The severity of the dental defects varies along the tooth row with later developing teeth most severely affected. Recent studies carried out on exfoliated (shed) deciduous