literature only four cases of trisomy 13 have been associated with imperforate anus: a de novo mosaicism 46,XX,t(13q13q)/46,XX,-13,+r(13), a partial trisomy of chromosome 13q and 20p and trisomy 13 with Robertsonian translocation of extra chromosome 13 to chromosome 14. The fourth case of trisomy 13 has been mentioned by Cho et al in 2001 in which the form of trisomy has not been described.

This baby girl was born at term by emergency caesarian section for fetal tachycardia to non consanguineous parents. On initial examination she was noted to have overlapping fingers, widely spaced nipples, ulnar deviation of hands, microphthalmia with ectropion of the right eye. Systemic examination revealed imperforate anus and a systolic murmur. The baby was transferred to a surgical unit and had a colostomy on day one of life under general anaesthesia. She was discharged home on day 21 of life.

Out of the four cases of trisomy 13 described so far with imperforate anus all have been either mosaics or translocations. Our baby is unique as she is possibly the only reported child with Patau syndrome 47 XX + 13 (pure non disjunction and not mosaic/translocation) to have anorectal malformation. It is still questionable whether imperforate anus is a true association of trisomy 13 or just a co-incidental occurrence.

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MOLECULAR TOOL IN THE DIAGNOSIS AND ANTENATAL DIAGNOSIS IN GLYCOGEN STORAGE DISEASE TYPE IA IN TUNISIAN PATIENTS

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Introduction: Glycogen storage disease type Ia is an autosomal recessive disorder of glycogen metabolism caused by glucose-6-phosphatase deficiency. It is characterized by short stature, hepatomegaly, hypoglycaemia, hyperuricemia, and hyperlactacidemia. The gene G6PC was cloned by lei et al in 1993.

Patients and methods: A study of the molecular basis of GSD type Ia was carried out in 8 Tunisian patients from 7 unrelated families. For five patients the diagnosis was performed by enzyme assay in fresh liver. To order to identify the mutations

segregating with Tunisian families, exons from G6PC gene were amplified for genomic DNA of patients and directly sequenced.

Results: Five patients had the R83C recurrent mutation and one had the R170Q. Both the two mutations are known to abolish G-6-Pase activity and therefore are responsible for severe form of GSD type Ia. R83C and R170Q are relatively frequent among Turkish patients.

Conclusion: High prevalence of the R83C mutation might be due not only to a hot spot mutation but also is probably due to population migration. As most of the Tunisian patients bear the R83C mutation, direct screening of this mutation will be a convenient alternative to the enzyme assay in fresh liver biopsy specimen for the diagnosis of GSD type Ia. Since this work all patients (22 cases) suspected of GSD type Ia were diagnosed by molecular biology.

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SLC25A19 MUTATION IS A NOVEL CAUSE OF NEUROPATHY AND BILATERAL STRIATAL NECROSIS

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Acute basal ganglia necrosis in the pediatric age group is a neurological disorder characterized by symmetrical degeneration of the caudate nucleus, putamen, and occasionally the globus pallidus. The outcome is less favorable for the familial cases where the course is relentlessly progressive or punctuated by recurrent episodes with involvement of additional brain regions and significant residual neurological damage. The differential diagnosis of the latter group includes mitochondrial respiratory chain defects organic acid disorders, Wilson disease, juvenile Huntington's chorea, pantothenate kinase-associated neurodegeneration, biotin responsive basal ganglia disease, and the striatal necrosis associated with NUP62 mutation.

In this study we report on four siblings from a consanguineous Arab Muslim family who suffered from recurrent episodes of flaccid paralysis and encephalopathy and chronic progressive

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