De novo mosaic ring chromosome 18 in a child with Behavioral Abnormalities And Epilepsy: Four Maternal age and trisomy 21

The phenotype of our patient and the role of TBX and NETOS-1 genes, involved in central nervous system myelination and aging of the lower limbs, is characterized by developmental delay/mental retardation, secondary microcephaly, typical dysmorphic features of 18q-, 18p-syndrome or a combination of both, intractable epilepsy, to our knowledge not described by now.

Aim: We report a case of de novo mosaic r(18) with a characteristic clinical presentation with the following features: micrognatia, dysmorphic ears, clubbing fingers and genital malformations. At the age of 10 he developed intractable myoclonic-astatic epilepsy that resulted in frequent falls and injuries. At present he is moderately mentally retarded, has clumsy gait and his somatic development follows 5th percentile for height and weight, while he is normocephalic.

Results: EEG recordings showed slower cerebral activity with diffuse paroxysmal discharges of high-voltage polysspike-wave complexes. MRI showed moderate dilation of lateral ventricles, small hyperintensive areas in peririgional white matter and depletion of white matter of dorsal corpus callosum. Standard karyotype revealed an excess of the Xp material attached to the terminal part of the 15q. FISH analysis performed by SHOX probe confirmed the excess of genetic material of Xp on 15q.

Conclusion: Our presentation is additional contribution to genetic cause of mental retardation, dysmorphic features and intractable epilepsy, to our knowledge not described by now.

Poster sessions S67

De novo mosaic ring chromosome 18 in a child with mental retardation, epilepsy and immunological problems.

A. Lo-Castro1 *, C. Galasso1, L. Di Carlo1, E. Souza Siqueira1 *, D. Bomfim2.

1Department of Neuroscience, Paediatric Neurology Unit, "Tor Vergata" University of Rome, Italy
2University of Brasilia, Brazil, 3Health center Zenica, Bosnia and Herzegovina, 4KB Zenica, Bosnia and Herzegovina

Ring chromosome 18 [r(18)] is a disorder in which one or both ends of chromosome 18 are lost and joined forming a ring-shaped figure. R(18) patients can therefore show features of 18q-, 18p-syndrome or a combination of both, depending on the size of the 18p and 18q deleted regions. The phenotype of the r(18) is characterized by developmental delay/mental retardation, secondary microcephaly, typical facial dysmorphisms and major abnormalities.

We report a case of de novo mosaic r(18) with a characteristic presentation of epilepsy by array based comparative genomic hybridization analysis. Our patient presents low intrauterine growth, dysmorphic features, short stature, hypothyroidism, celiac disease, ptotic right kidney, mitralic insufficiency, and focal seizures.

A neurocognitive assessment showed a mental retardation of mild-moderate degree, attention and visuo-spatial deficits, and hypoactivity. Awake EEG was characterized by isolated, synchronous and asynchronous spikes on posterior regions. Brain MRI showed slight hyperintensities in semiovular areas, at the level of frontal subcortical white matter. After contrast application, these foci did not show potentiation, suggesting non-specific gliotic lesions.

The phenotype of our patient and the role of TBX and NETOS-1 genes, involved in central nervous system myelination and plasticity, are discussed.

Maternal age and trisomy 21

E.G. Gvozdenović1 *, S. Kadiric2, L. Hukeljčiči3. 1Cantonal Hospital Children's Department, Bosnia and Herzegovina, 2Health center Zenica, Bosnia and Herzegovina, 3KB Zenica, Bosnia and Herzegovina

Syndrome Down is the most common chromosomal disorder. Incidence among live-born children is about 1:750. Incidence of trisomy 21 increases proportionately with increasing maternal age. Although the risk in younger women is significantly smaller, they constitute about half of the total number, due to multiple pregnancies at that age.

Aim: was to present average age of the mothers and age group with the highest incidence of disease. We made retrospective analysis assessing mothers of children with Down syndrome, born at Obstetrics Department in Zenica. Thirty-six children with clinical signs of trisomy 21 were born in period 1999-2009. We analyzed the following factors: maternal age, sex, gestational age, parity, associated anomalies.

Results: showed statistically significant number of mothers of children with this syndrome in the age group under the 30 years: 5.5% of mothers were younger than 20 years; 50% aged 20–30 years. However 25% were aged 30–40 years and 19.5% were older than 40 years. Small number of mothers in the later childbearing may be explained by the perpetration of prenatal diagnosis and medically indicated abortion.

Conclusion: all pregnant women, regardless of age, must have access to screening for Down syndrome in the second trimester of pregnancy using serum tests (free β-hCG, unconjugated estriol, α-fetoprotein) and ultrasonography. Amniocentesis is a method of choice for mothers older than 35 years.
Conclusion: The association between XYY syndrome, multiple hereditary exostoses and epilepsy has not to our knowledge been described. This case also shows the need for more studies on the eletroclinical traits of epilepsy in children with XYY syndrome.

**P08.10** Klinefelter syndrome and other sex-chromosomal abnormalities

I. Hsairi1,*, H. Ben Othmen2, N. Belghith1, E. Ellouz1, F. Kamoun1, H. Kamoun3, C. Triki1. 1Department of child neurology research unit neuropediatric, Tunisia, 2Research unit neuropediatric; 3Laboratory of Molecular Genetics, Department of genetics, Tunisia

Background: A great number of sex-chromosomal abnormalities (SCA) have been detected associated with mental retardation (MR). They are expressed phenotypically in different kinds of syndromes: Klinefelter, Turner’s syndrome...

Methods: A prospective study was conducted on 270 children with MR. They were subjected to a detailed family history, physical examination with photographs. Karyotype analysis was done and genomic DNA was extracted in all cases.

Results: SCA were finding in four children with mild MR. Two male were diagnosed as KF syndrome. The first one, 11 years old, was followed for learning disabilities. Her clinical examination was normal. The second case, 2 years old child, had facial dysmorphism, hypotonia and ambiguous genitalia. The karyotype confirmed respectively the diagnosis of classic KS and KS variant. Mutations in MeCP2 have been screened.

In the third case, a 11 years old girl was followed for learning disability, short stature, and macroglossia. The karyotype analysis performed on mild mentally retarded children especially in developing MS, is discussed.

Conclusion: Screening for SCA by karyotype should be performed on mild mentally retarded children especially those with dysmorphic or genital abnormalities. We stressed on a multidisciplinary approach in order to reach prompt diagnosis, best follow up and focus on X-linked gene in some SCA.

**P08.11** Macrocephaly, dysmorphic features, West syndrome and Mental Motor Retardation due to unbalanced segregation of familial reciprocal translocation between chromosomes 8 and 9

I. Erol1, S. Saygi1,*, F. Alehan1, F. Sahn2. 1Department of pediatrics, Division of Child Neurology, Baskent University Faculty of Medicine, Turkey, 2Department of Medical Genetics, Baskent University Faculty of Medicine, Turkey

NR3C2 or mineralocorticoid receptor mutations are the principal cause of autosomal dominant or sporadic type 1 pseudohypoaldosteronism. Patients can be asymptomatic or show a salt-losing syndrome in the neonatal period. Afterwards, patients compensate for their defective mineralocorticoid receptors by upregulating their mineralocorticoid axis, hence presenting, in most cases, a lifelong increase in renin and aldosterone levels.

The renin–angiotensin–aldosterone system (RAAS) is a key hormonal system regulating blood pressure by acting on vasculature, heart, kidney and adrenal gland. However, expression of RAAS components has recently been detected in other tissues such as brain, thymus and immune cells. Moreover, the RAAS has been implicated in several mouse models of autoimmune disease. Series of studies have demonstrated that angiotensin II has a wide variety of proinflammatory properties which may contribute to the development of autoimmune diseases. Notably, angiotensin II seems to play a role in sustaining autoimmune neuroinflammation in chronic experimental autoimmune encephalomyelitis, a model mimicking many aspects of multiple sclerosis (MS), and angiotensin receptors are upregulated in infiltrates of plaques from the brains of MS patients.

We report on a male patient who presented at the age of 12 years with MS and in whom an inherited microdeletion within the NR3C2 gene was detected, compatible with the diagnosis of type 1 pseudohypoaldosteronism. His mother and her sister, who both developed autoimmune thyroiditis, also carry the microdeletion. The clinical relevance of this microdeletion in developing autoimmune diseases, and especially in developing MS, is discussed.
学霸图书馆（www.xuebalib.com）是一个“整合众多图书馆数据库资源，提供一站式文献检索和下载服务”的24小时在线不限IP图书馆。图书馆致力于便利、促进学习与科研，提供最强文献下载服务。