

Selected Abstracts of the 13th International Workshop on Neonatology

THE POWER OF EPIGENETICS TWINS: IDENTICAL BUT DIFFERENT

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The Workshop has been organized with the patronage of the Italian Society of Neonatology (SIN), the Italian Society of Pediatrics (SIP), the Italian DOHaD (Developmental Origins of Health and Disease) Society, the Italian Society of Preventive and Social Pediatrics (SIPPS), the Union of European Neonatal and Perinatal Societies (UENPS), the International Federation of Clinical Chemistry and Laboratory Medicine (IFCC), the Union of Middle-Eastern and Mediterranean Pediatric Societies (UMEMPS), the European Association of Perinatal Medicine (EAPM) and lastly the Italian-Romanian Society of Pediatrics.

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any definitive conclusion on the role of maternal nutrition and/or occupational and living areas on their concentration. Larger studies are needed to determine these aspects and even to correlate any possible positive or negative effect on newborn growth and development. In particular mother living in mining areas should be monitored as the infants could be at risk of multiple exposure to mining related toxic metals, such as lead, mercury, cadmium and arsenic, through breast milk intake, in addition to *in utero* exposure.

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METABOLOMIC ANALYSIS OF PLACENTA TISSUE IN OBESE PREGNANCIES

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INTRODUCTION

The placenta plays a pivotal role as it represents the connection between mother and foetus. It acts as a carrier (nutrients and oxygen, waste products and carbonic anhydride) as well as a barrier against infection. Pregnancy related metabolomics has examined different biofluids in order to reach deeper insights into pregnancy pathologies [1]. Amniotic fluid, urine and plasma were the most studied, while placenta tissue has been the subject of few studies [2]. In this report, a method to analyse the placental metabolome through the GC-MS (Gas Chromatography-Mass Spectrometry) technique is described.

MATERIALS AND METHODS

Placenta tissues were collected after elective caesarean section from obese and normal weight mothers, snap frozen and stored at -80°C until analysis. Each sample was homogenized with a

mixture of solvents (chloroform, methanol, water), centrifuged, and the obtained phases were separated and processed following two different protocols. The hydrophilic phase was vacuum dried and treated with two derivatizing reagents, methoxyamine hydrochloride and MSTFA (N-Methyl-N-[trimethylsilyl] trifluoroacetamide), to give a mixture of trimethylsilylated metabolites. The lipophilic phase was dried and treated with boron trifluoride in methanol to obtain a mixture of FAME (Fatty Acid Methyl Esters). The two phases were separately analysed on a GC-MS platform, applying different instrumental conditions. The chromatograms obtained were analysed with the free software AMDIS (Automated Mass Spectral Deconvolution and Identification System) using a lab-made library comprising 200 metabolites.

RESULTS

The analysis of the placenta tissue allowed to identify essential metabolites. In the hydrophilic fraction, 78 metabolites were detected: they mainly belong to the classes of amino acids (18), carboxylic acids (13), sugars and sugar-related compounds (11), phosphorylated derivatives (9). The lipophilic fraction analysis allowed to define the fatty acid profile of the placenta extract: the saturated fatty acids palmitic and stearic, the mono-unsaturated oleic, and the polyunsaturated (PUFA) linoleic, arachidonic, 8,11,14-eicosatrienoic and 4,7,10,13,16,19-docosahexaenoic acid (DHA) were the most representative. Statistical analysis of the data matrices derived from the hydrophilic and lipophilic fraction analysis were performed in MetaboAnalyst 3.0 [3], to investigate the possibility of identifying characteristic metabolic differences between the different phenotypes. Placentae from obese mothers were compared with those from normal weight mothers to get new insights into the correlation between the metabolite content and metabolic status.

CONCLUSIONS

The reported analysis method allowed to examine different metabolites in placenta samples, divided into two phases, hydrophilic and lipophilic. All metabolites were analysed through the same GC-MS platform to provide interesting informations about the placenta tissue composition.

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PATAU SYNDROME: RADIOLOGICAL CHARACTERISTICS

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INTRODUCTION

Patau syndrome (Trisomy 13) is a rare chromosomal anomaly caused by the presence of an extra chromosome 13 and characterized by multiple somatic and visceral malformations, associated with severe mental impairment. Its incidence is estimated around 1:16,000 newborns and only 5 to 10 percent of children with this condition live past their first year. Postpartum diagnosis of trisomy 13 has been drastically reduced by the prenatal screening tests.

CASE REPORT

We're presenting the case of a patient with multiple malformations, who was diagnosed with complete trisomy 13 (karyotype: 47 XY +13). An augmented risk of chromosomopathy was found during first semester screening tests and confirmed by fetal US scan and MRI. Nevertheless, parents refused to undergo any invasive procedure to determine the disease. At birth the little patient presented scaphocephaly with patent bregmatic fontanelle, microphthalmia and micrognathia. Both hands had a subcentimetric appendage attached to the ulnar side and the right foot showed hexadactyly with syndactyly of V-VI toe. Bilateral cryptorchidism was also noted. Pre and postnatal US scans evidenced hypoplasia of the left kidney and malrotation of the right one, persistence of the Botallo duct and a suspect of interatrial defect with left to right shunt. MRI showed cerebral pachygyria and absence of both crystalline lenses. The clinical conditions were critical from birth, since the patient presented respiratory distress, and worsen at the third day of age, when the patient developed a gastric perforation and underwent an emergency surgery. After several episodes of desaturation, the little patient died at day 21.

CONCLUSIONS

The aim of this case report is to recall the clinical and radiological characteristics of this rare genetic syndrome, since the postnatal diagnosis is becoming unusual in our daily practice.

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A CASE REPORT OF BECKWITH-WIEDEMANN SYNDROME: THE LENS OF THE RADIOLOGIST

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INTRODUCTION

Beckwith-Wiedemann Syndrome (BWS) is a pediatric overgrowth disorder, with an increased risk of childhood tumors. The incidence of BWS is estimated to be 1 out of 13,700 and is caused by various epigenetic and/or genetic alterations. It is characterized by many additional clinical features (major criteria) such as hypoglycemia, macrosomia, macroglossia, hemihyperplasia, cardiomyopathy, omphalocele, visceromegaly, adrenocortical cytomegaly, renal abnormalities, etc. The diagnosis of BWS is established with three major criteria or an epigenetic or genomic alteration leading to abnormal methylation at 11p15.5. Molecular genetic testing for this alteration can be done also on amniocytes in prenatal test (in high risk pregnancies due to positive family history) as well as ultrasound examinations to assess fetal growth and to detect other abnormalities characteristic of BWS (no high risk pregnancies). Most important differential diagnosis with other overgrowth syndromes like: Costello, Sotos, SGB type I. Thanks to the recent improvements in syndrome recognition and treatment the overall mortality rate of BWS is about 10-20% with most deaths occurring early secondary to congenital malformations or prematurity.

CASE REPORT

A preterm (27 w and 1.4 kg) female, born by caesarean section, showed omphalocele, right emihypertrophy and respiratory distress (also seen on first X-ray). Ematochemical analysis showed: hypoglycemia, increase of bilirubin (tot 10 mg/dl), all cholestasis index (GGT 190 U/I, LDH 1,085 U/I), CPK (682 U/I) and WBC (12,000/μl). Suspecting other organs abnormalities, a study of abdomen with Ultrasound (US) was required. US showed an important hepatomegaly (right lobe 77 mm), bilateral dishomogeneous hypoechoic mass in