PP-042

Association of an increased nuchal translucency with total agenesia of the ductus venosus

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Looking for the etiology of an increased nuchal translucency in a patient aged of 22 years G1P0, We performed initially a karyotype whose formula was normal: 46xy. A Fetal echocardiography realized at 18 and 22 weeks' gestation was normal and brain MRI was normal also. At 22 weeks' gestation, ultrasonography revealed a hepatic vascular abnormality without other symptoms. The suspected diagnosis was agenesis of the ductus venosus with creation of a high grade arteriovenous shunt between the umbilical vein and the inferior vena cava. The geneticist suspected also a genetic syndrome but the patient refused any investigation. Agenesis of the ductus venosus may be one expression of the different possible systemic-portal-umbilical abnormalities. Physiological consequences vary according to the type of substitutive anastomoses. Agenesis of the ductus venosus can be strictly isolated. It can be also one of a manifestation of a genetic syndrome like NOONAN and JOUBERT SYNDROME. In our case, agenesia of the ductus venosus was associated to an increased nuchal translucency. We did not find a similar case in the literature.

Keywords: Nucal translucency, arantius.

PP-043

Congenital cataract as an only sign in Down Syndome: case report

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Case: 42 year old pregnant woman has been sent to perinatology policlinic for double test. Her first trimester combined test result was 1/1240. She was appointed for second trimester detailed sonography for advanced maternal age. Bilateral opacities in orbital examination were seen on sonography. She was given genetic consultation for this but she opted to continue her pregnancy without karyotyping. Planned c/s section was done for previous c/s endication at 39th week. A female 3450 gr fetüs was born with an Apgar score 8-9. Her face was characteristic with upward slanting of the palpebral fissures, congenital cataract and macroglosi. Postnatal karyotype was concordant with Down syndome.

Conclusion: In high risk groups orbital examination should be included in prenatal sonography.

Keywords: Down syndome, eye, cataract.

PP-044

Ellis-Van Creveld syndrome

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Objective: To present a prenatally diagnosed case of Ellis-Van Creveld (EVC) syndrome and discussion of management for this case.

Case: A 19 years old gravida 1, para 0 was referred to our unit because of fetus with shortness in all long bones at 21 weeks gestation. Parents had consanguineous marriage. A detailed ultrasound scan revealed a single live fetus with shortness of long bones (<2.5 th) severe thoracic hypoplasia with short ribs, postaxial polydactyly of bileteral hands, and aortic hypoplasia with atrioventricular septal defect.After counseling about the fetus and prognosis, cordosentesis was performed and the pregnancy was terminated. Postmortem examination confirmed as a Ellis-Van Creveld Syndrome. Karyotype analyse was revealed a 46,XX karyotype of the fetus.

Conclusion: EVC syndrome can be diagnosed by prenatal sonograpy in the second trimester but EVC should be distinguished from thoracic asphyxiating dystrophy (Jeune syndrome) and the group of short-rib polydactyly syndromes.

Keywords: Ellis-Van Creveld syndrome, prenatal diagnosis.

PP-045

Early neonatal outcomes of idiopathic polyhdramnios

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Objective: Polyhdramnios is seen nearly 0.4%-%1.7 of the obstetric population after 24th week. About 45% of these cases etiology can not be defined. Nearly 5% of the idiopathic polyhdramios post natal structural anomaly is detected. Gastro intestinsl system, urinar system and cardiovascular system anomalies are the most seen ones. Risk of having aneuploidy is higher in idiopathic polyhdramnios. Stuides have showed that early neonatal outcomes is not well when compared with