FCP156

DENTES NATALES: OLGU SUNUMU

***Kılınç N., **Demir B., ***Yavuz İ., **Yayla M.,** *Dicle Üniversitesi Tıp Fakültesi *Patoloji A.D., **Kadın Hastalıkları ve Doğum A.D., ***Diş Hekimliği Fakültesi Pedodonti A.D., Diyarbakır - Türkiye*

Dentisyon (dişlenme) doğumdan sonra 6. ayda alt orta süt keserlerin çıkması ile başlar. 12000 canlı doğumda bir görülen intrauterin çıkan dişlere dentes natales denmektedir. 22 yaşında G3, P2, Y0 olan olgu polihidroamniyos tanısı ile perinatoloji polikliniğine başvurdu. Yapılan ultrasonografide fetusun her iki bacağında hiperekstansiyon mevcuttu. Spontan kontraksiyonları başlayan anne gebeliğinin 36. haftasında 1650 gr ağırlığında, 47 cm boyunda 1/0 Apgar skorlu bir kız bebek doğurdu. Otopsi incelemesinde, burun kökü basık, kısa boyun, üst süt keserler bölgesinde iki adet kesici formunda kıkırdağımsı yumuşak, sağ alt süt I nolu kesici diş bölgesinde bir adet süt santral formunda sert diş, bacaklarda hiperektansiyon ve pes planovarus (vertikal talus) saptandı. Malformasyonlar ile birlikte üç adet natal diş tesbit edilen olgu nadir görülmesi nedeniyle sunulmaya değer bulundu.

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ANTENATAL DIAGNOSIS OF RING CHROMOSOME 22

Ayhan C.*, Apaydın H.*, Öncüloğlu C.*, Kılıç G., Çolgar U.*,** **Acıbadem Hastanesi Kadın Doğum Kliniği, **Acibadem Hastanesi Genetik Labaratuvarı, İstanbul - Türkiye*

Ring malformation of chromosome 22 is an extremely rare disease . The diagnosis usually achived by caryotype analysis during early childhood period. Indications of caryotype analysis are mental retardation, brain tumors, language delays and face-head malformations of the babies such as hipotelorism, dolychocephaly.

We are presenting a case which was diagnosed at as early as 16th week of pregnancy. Mrs.OS. admitted to our clinic for routine antenatal examination on her 6th week of pregnancy. Her medical history was unremarable and initial lab. tests were with in normal range including TORCHES. She diagnosed as hyperemesis gravidarum at 9th week and treated with iv fluids because she lost 5% of her prepregnant weight. On her first trimestr usg screening , nuchal translucency measured as 2.1 mm and there was not any accompining malformation. The size of the baby was exactly as it supposed to be according to LMP. Triple screening results of this patient were negative for trisomy 21 (1/330), trisomy 18 (1/10000) and NTD (1/10000)at the 16th week. The only problem was relatively increased trisomy 21 incidence compared to age risk and extremely high levels of hCG (5.1 MoM). At this time she had second level ultrasonography examination and the finding were as follows: Shape of the head was mimicing lemon sign, BPD was shorter then where HC(head circumference) was in normal range and FL was shorther for two weeks. At this point she was suggested to take antenatal caryotype analysis and the result reported as 46, XX, r(22)[65] / 45,XX,-22[7] / 47,XX,r(22),r(22)[3]. In oder to eliminate maternal dissemination patient and the father of the baby get karyotype analysisis and the results were normal. Ultrasonography performed at the 20th week and we have found hyperechogenic bilateral kidneys, hipotelorism, dolychocepahaly, unilateral choroid plexus cyst and significant growth retardation of limbs.

Patient get genetic counselling and decided to terminate the pregnancy since 100% mental retardation is expected with this chromosomal abnormality.

We recommed antenatal caryotype analysis in every patient whose NT levels over 2 mm with the presence of triple screening deviations. We conclude that NT measurement is an essential part of pregnancy follow-up which lead us to diagnose rare abnormalities such as the case presented. .If there is any accompining usg finding and /or triple screening deviation antenatal caryotype analysis must be considered even NT measurements with in normal range.

Ring chromosome 22 is a rare reason of mental retardation. The antenatal findings of the disease ar early growth retardation of fetal limbs, asymmetric shape of head and increased nuchal translucency Extremely high levels of hCG should be considered for caryotype analysis.