

EP10.04

Agensis of peroné: a case reportJ. Fernández Merino², I. Javier¹, N. Cornejo¹, C. Rojas¹, N. Lagos¹, C. Cabrera¹¹Hospital de Carabineros de Chile, Santiago, Chile;²Ginecología y Obstetricia, Hospital de Carabinero de Chile, Santiago, Chile

Malformations of the lower extremities are rare and poorly described. Although fibular agenesis is the most common lower extremity malformation, there are few published cases of prenatal diagnosis. We report a clinical case of fibular agenesis that was presented at the Hospital de Carabineros de Chile and its subsequent discussion.

Supporting information can be found in the online version of this abstract

EP10.05

Relationship between absent or hypoplastic fetal nasal bone at 20–23⁺6 weeks of gestation and chromosomal defects in an unselected Chinese population

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Objectives: To investigate the relationship between absent or hypoplastic fetal nasal bone and chromosomal defects during the second trimester in an unselected Chinese population.

Methods: From January 2012 to December 2015, included were 58133 pregnant women who underwent routine ultrasound examination between 20 and 23+6 gestational weeks as a screening test. We collected all the cases of absent or hypoplastic fetal nasal bone, followed up the results of karyotype analysis and the information on neonatal development.

Results: (1)150 fetuses were found to have absent or hypoplastic fetal nasal bone in the 58651 fetuses(2.56%), of which 57636 were singleton,476 were twins and 21 were triplets. With 14 cases of misdiagnosis, missing or incomplete data excluded,136 cases were included in the study. 113 underwent interventional prenatal diagnostic test, and fetal chromosomal defects were detected in 24 cases (17.6%), including 18 cases (75%) of Trisomy 21, 3 cases (12.5%) of Trisomy 18, 1 case (4.2%) of Klinefelter syndrome and 2 cases (8.3%) of microdeletion syndrome. (2) There was no significant difference in the incidence of chromosomal defects between absent fetal nasal bone and hypoplastic fetal nasal bone[22.5%(16/71) vs 12.3%(8/65), $\chi^2=2.442$, $P=0.118$]. (3) The incidence of chromosome defects in fetuses without other structural defects was significantly lower than that with structural defects [3.9%(3/76) vs 35.0% (21/60), corrected $\chi^2=22.247$, $P=0.000$]. (4)A total of 38 cases of Down syndrome were found in 56707 cases of delivery or induced labour in our hospital. When the fetal nasal bone dysplasia was used as an indicator of Down's syndrome, the sensitivity was 47% and the specificity was 99.8%.

Conclusions: The absent and hypoplastic fetal nasal bone are closely related to fetal chromosomal defects. When combined with other sonographic defects, it is necessary to carry out a detailed prenatal diagnosis to exclude fetal chromosomal defects.

EP10.06

HD-flow use in the study of the posterior palate defects: pilot studyA. Comanescu¹, N. Cernea², F. Tanase⁴, O. Tica³, M. Barbu⁴, C. Comanescu⁴¹SCJU Craiova, Craiova, Romania; ²Obstetrics and Gynecology, University of Medicine and Pharmacy, Craiova, Romania; ³Mother and Child, University of Medicine and Pharmacy Craiova, Craiova, Romania; ⁴Obstetrics and Gynecology, University Hospital Craiova, Craiova, Romania

Objectives: In our routine examinations we noticed that we could follow fetal swallowing using colour Doppler - we tried to study if this technique could be used to monitor the contour of the fetal palate in order to screen for posterior palate defects.

Methods: We shared the idea with our team in the Prenatal Diagnosis Unit and we monitored the accessibility of HD - Flow in assessing the contour of the fetal palate on a fetal profile section. From 27.09.2016 to 23.03.2017 we assessed 325 patients using this technique.

Results: Out of 325 cases enrolled, the images were successfully obtained in 314 cases. There was one case of posterior palate defect with associated micrognathia, that was suspected on 2D and also on the HD-flow scan. Five out of six examiners considered that using the HD-Flow increased their confidence that the posterior palate was normal.

Conclusions: There are other methods of assessing posterior palate, using 2D or 3D scans. Our methods come only to complete these types of examinations and to increase the confidence of the diagnosis.

EP10.07

Abstract withdrawn

EP10.08

Look at the fetal nose 1: the nasofrontal angleE. de Jong-Pleij^{1,2}, C.M. Bilardo³, G. Manten²¹Obstetrics, St Antonius Hospital, Utrecht, Netherlands;²Woman and Baby, University Medical Centre Utrecht, Utrecht, Netherlands; ³Fetal Medicine, University Medical Centre, Groningen, Netherlands

Objectives: To study with 3D ultrasound the fetal nasofrontal angle (NFA) in 2nd and 3rd trimester fetuses and assess reproducibility, construct normal ranges and compare these results with pathological cases.

Methods: The NFA, defined as the angle between the nasal and the frontal bones, was measured in the exact mid-sagittal plane of the fetal profile in 3D volumes. The mean of two measurements was used. 109 healthy Caucasian fetuses and 16 cases with a pathological condition known to be associated with deviant facial features (thanatophoric dysplasia, Apert syndrome, achondroplasia, hydrocephaly, Trisomy 13, lissencephaly, Nager syndrome, maxillonasal dysplasia, chondrodysplasia punctate, frontonasal dysplasia, Stickler syndrome, Pallister Killian syndrome, campomelic dysplasia, Cornelia de Lange syndrome, Cri-du-chat syndrome and Wolf-Hirschhorn syndrome) were analysed.

Results: The ICC for inter- and intraobserver reproducibility was 0.86 and 0.95, respectively. Between 16 and 35 3/7 weeks the NFA did not change significantly with a mean of 117° (10th and 90th centile, were 105° and 129° respectively). In all 4 cases with frontal bossing (thanatophoric dysplasia, Apert syndrome, achondroplasia, hydrocephaly) the NFA was below the 10th centile. In all 3 cases with microcephaly (Trisomy 13, lissencephaly, Nager syndrome) and in maxillonasal dysplasia, chondrodysplasia punctate, frontonasal

dysplasia and Wolf-Hirschhorn syndrome the NFA was above the 90th centile. In cases with Stickler syndrome, Pallister Killian syndrome, campomelic dysplasia, Cornelia de Lange syndrome and Cri-du-chat syndrome the NFA was within the normal range.

Conclusions: The NFA is stable in the 2nd and 3rd trimester with a mean of 117°. The NFA can support subjective evaluation of the fetal profile and be of help in classifying and diagnosing fetuses with nasal anomalies (especially flat noses) and forehead anomalies.

EP10.09

Added value of prenatal MR in the evaluation of fetal cleft lip, cleft palate and micrognathia

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Objectives: The aim of this study was to describe MR findings and to determine the accuracy of fetal MRI in diagnosing fetal cleft lip, cleft palate, and micrognathia, either isolated, or in association with additional abnormalities.

Methods: We reviewed our fetal MRI database from March 2011 through February 2017 and searched for exams referred for cleft lip, cleft palate, and/or micrognathia. A total of 37 fetal exams were identified. 7 cases were lost to follow up, 6 patients underwent termination of pregnancy. Anatomic facial landmarks and biometric parameters were documented including evaluation of the lips, maxillary ridge, position of the tongue, presence/absence of the palate, measurement of the jaw index to evaluate for micrognathia, and binocular and intraocular diameter to evaluate for hypo/hypertelorism. A complete fetal neuroevaluation was performed to detect additional anomalies. Findings on fetal MR were correlated post delivery.

Results: In the remaining cohort of 21 fetuses, 20 cases were diagnosed with positive findings. There were 6 diagnoses of cleft lip, 7 diagnoses of cleft lip and palate, 4 isolated micrognathia, 5 micrognathia with isolated cleft palate consistent with Pierre Robin sequence. Of the 21 fetuses, the MR diagnosis was confirmed following delivery in 17 cases. Syndromes that were seen with micrognathia included Dandy Walker, Frynn's, Emanuel, and mid-face hypoplasia syndromes. There was 1 false positive case of cleft lip and palate due to a tethered frenulum.

Three cases are waiting to deliver.

Conclusions: MRI has a 95.5% degree of accuracy in diagnosing fetal facial abnormalities, including isolated cleft lip, cleft lip with cleft palate, micrognathia with cleft palate (Pierre Robin syndrome), and isolated syndromic micrognathia with additional non-palatal abnormalities.

EP10.10

Look at the fetal nose 2: the nasal height

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Objectives: To study with 3D ultrasound the reproducibility of fetal nasal height (NH) in 2nd and 3rd trimester fetuses, create normal ranges and compare these results with pathological cases.

Methods: After multiplanar correction to the exact mid-sagittal plane the head position was standardised by turning the fetal profile line exactly vertical (aligning the lower part of the forehead above the anterior part of the mandible). Nasal height, defined as the

vertical nasion (bone) – subnasale (skin) distance, was measured with the 'distance between two lines' option. The mean of two measurements was used. 109 healthy Caucasian fetuses and 16 cases with a pathological condition known to be associated with deviant facial features (thanatophoric dysplasia, Apert syndrome, achondroplasia, hydrocephaly, trisomy13, microcephaly, Nager syndrome, maxillonasal dysplasia, chondrodysplasia punctate, frontonasal dysplasia, Stickler syndrome, Pallister Killian syndrome, campomelic dysplasia, Cornelia de Lange syndrome, cri du chat syndrome) were analysed.

Results: The ICC for inter- and intraobserver reproducibility was 0.96 and 0.95, respectively. NH increased between 16 and 35 3/7 week from 7.3 to 18.6 mm (NH=11.384 + 0.204GA – 0.000335GA², r²=0.86 (GA=gestational age in days)). NH was below the 10th centile in cases with thanatophoric dysplasia, Stickler syndrome, Pallister Killian syndrome, campomelic dysplasia and cri du chat syndrome. The NH was within the normal range in the other 11 cases.

Conclusions: NH increases between 16 and 35 3/7 weeks from 7.3 to 18.6 mm. The NH can support subjective evaluation of nasal abnormalities and be of help in recognising the phenotype of a specific syndrome.

EP10.11

The frontal fetal facial (FFF) angle in the second trimester measured by 3D ultrasound in normal fetuses and fetuses with Trisomy 18

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Objectives: The aim of this prospective study was to establish reference bands for the FFF angle (forehead-nose-mandible) in normal second trimester fetuses and to compare the normal data with the FFF measurements of fetuses with confirmed Trisomy 18.

Methods: In 294 normal fetuses and 21 fetuses with Trisomy 18 the FFF angle was measured in the multiplanar 3D mode after aligning the fetal head into an exact upright position to get the true profile. All scans were performed using E8/E10 GE equipment (Zipf, Austria) with a 5-8 MHz 3D abdominal or a 5-9 MHz 3D vaginal transducer. Gestational age was between 14+0 and 27+0 weeks of gestation.

Results: In the normal fetuses the 90% reference band for the FFF angle was calculated. The upper limit of normal shows a value of 151° at 14+0 weeks' gestation and 149° at 27+0 week's gestation. In 10 of the 294 normal cases (3.4%) the FFF angle was measured above the normal range and in 9 cases (3.1%) below the lower limit. In the 21 Trisomy 18 fetuses only 5 (23.8%) of them had a FFF angle above the upper limit while 15 (71.4%) of them showed values within the normal range and in 1 case (4.8%) the angle was measured below the lower limit.

Conclusions: In most of the Trisomy 18 cases the FFF angle was within the normal range, i.e. most of them did not show a flat profile. Thus the FFF angle is not a useful single parameter to detect Trisomy 18 fetuses in the second trimester.