

Mean z-score, 95% CI and centile corresponding to the mean were calculated. One-sample t tests were carried out to verify if z-score distribution is equal to zero. Finally, we compared z-scores of male and female fetuses with Down syndrome (DS) by the means of two sample t test.

Results: 132 CHA pregnancies was eligible for designed analysis, of which 80 cases with DS, 20 with Edwards syndrome (ES), 7 with Patau syndrome (PS), 17 with Turner syndrome (TS) and 8 with triploidy. The results are summarized in table. We proved no difference in growth pattern of fetuses with DS. The separate comparison of female and male fetuses with DS also revealed no significant difference ($p>0,2$). Otherwise, we observed significant growth restriction – similar in PS and TS (around 5th centile), severe in ES (0,5th centile) and the most striking in triploidies (<0,003th centile).

Conclusions: Statistical analyses showed no significant difference in growth pattern of DS fetuses, regardless of their sex. By contrast, in TS, PS, ES and triploidy there was growth restriction of various extent, deepening in mentioned order.

OP08.02

Crown-rump length in genetically abnormal pregnancies compared to a CRL curve for normal pregnancies

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Objective: In the first trimester of pregnancy, some aneuploidies have been reported to show a smaller size for gestational age (GA). The aim of this study was to compare crown-rump length (CRL) in pregnancies with a genetic abnormality with a new reference curve derived from normal pregnancies between 5.5 and 14 weeks GA.

Methods: A retrospective database study of the CRL of embryos and fetuses at different gestations. A CRL curve for normal pregnancies was obtained using a linear mixed-effects analysis. Subsequently the deviation from the CRL in pregnancies with a chromosomal abnormality was calculated and expressed as a z-score.

Results: A linear mixed-effects analysis was previously performed on 3710 normal pregnancies. Z-scores were calculated for all 4387 data points, with an internal validation on 30% of the data. A one sample t test was then performed on 155 scans from 134 genetically abnormal singleton pregnancies. The mean of the z-scores of the CRL of these genetically abnormal singleton pregnancies was $-0.109 [-1.462\ 0.655]$. This is statistically significantly lower than the expected CRL value at a specific GA for normal pregnancies. For Trisomy 18 pregnancies, a median z-score for the CRL of $-1.197 [-2.914\ 0.093]$ with $p<0.0001$ was observed.

Discussion: Pregnancies with some genetic abnormalities show a smaller than expected CRL when compared to a reference CRL

curve of normal singleton pregnancies. The difference in CRL is most pronounced for trisomy 18 pregnancies.

OP08.03

Chromosomal anomalies spectrum in missed miscarriages in relation to embryo appearance

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Objective: To compare the cytogenetic findings in a series of missed miscarriages studied by chorionic villus sampling, in relation to embryo appearance (embryonic or anembryonic).

Material and Methods: From January 2002 to January 2008, transcervical chorionic villus sampling (TC-CVS) was offered to women presenting with a missed miscarriage, for karyotyping. Previously, a transvaginal ultrasound examination was performed to assess embryo presence or absence, the CRL, and sac measurements. Cytogenetic analysis of the chorionic villi was carried out by means of the semi-direct method.

Results: Among 257 missed miscarriages with TC-CVS, the karyotype was obtained in 180 cases (70%), in 71% when the embryo was present (138/193) and in 65% (42/64) in miscarriages with an apparently empty sac. No differences in maternal age were found between both groups (35 ± 5 vs. 36 ± 5). A chromosomal abnormality was found in 66% (n=92) of miscarriages with an apparent embryo and in 59% (n=17) of those with an empty sac. The most frequent anomalies in miscarriages with an apparent embryo were trisomy 21 (13 cases) and monosomy X (13 cases), whereas trisomy 16 (8 cases) was the most frequent in those with an empty sac. Interestingly enough, no trisomies 21,18 nor monosomies X were found in the latter.

Conclusions: Viable aneuploidies were more frequently found in missed miscarriages with an apparent embryo, whereas uncommon autosomal trisomies were more common in miscarriages with an empty sac.

OP08.04

Increased risk of cleft lip/palate in case of enlarged NT

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Objective: The aim of this study was to evaluate whether in chromosomally normal fetuses there is an association between an enlarged nuchal translucency (NT) and isolated cleft lip/palate.

Methods: Pregnancy outcome of all women who underwent a NT measurement at our Prenatal Diagnosis Unit between January 2000 and November 2007 was reviewed.

OP08.01: Table

Chromosomal syndrome	Number of cases	Mean z-score	95% CI	Centile corresponding to mean	p value (one sample t test)
Down syndrome	80	0.22	(-0,18; 0,63)	58.4	$p>0.2$
Edwards syndrome	20	-2.55	(-3,1; -1,99)	0.5	$p<0.001$
Patau syndrome	7	-1.74	(-3,03; -0,44)	4.1	$p<0.02$
Turner syndrome	17	-1.63	(-2,28; -0,98)	5.2	$p<0.001$
Triploidy	8	-4.3	(-6,35; -2,26)	<0,003	$p<0.01$