Typical structural abnormalities of trisomy 13 detectable by ultrasound during first trimester screening

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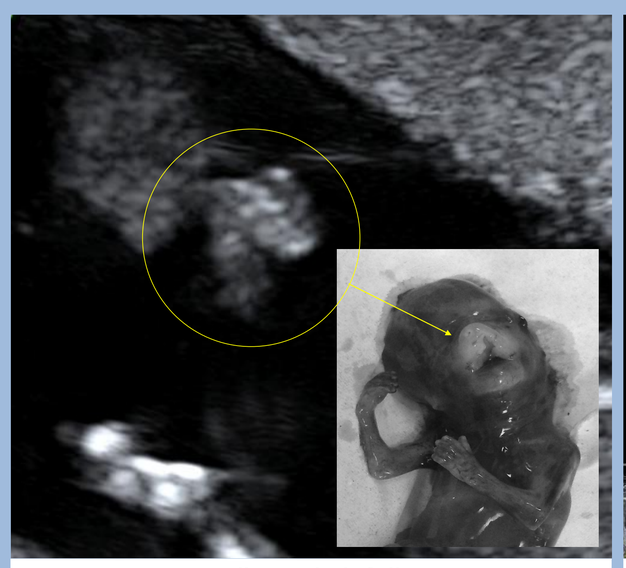
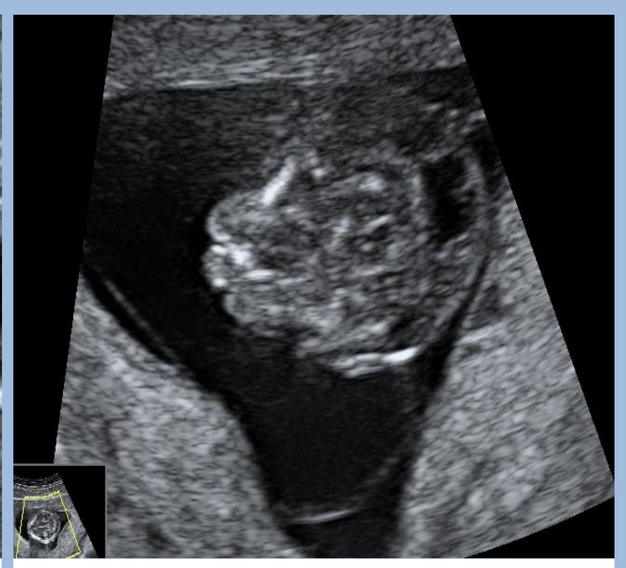
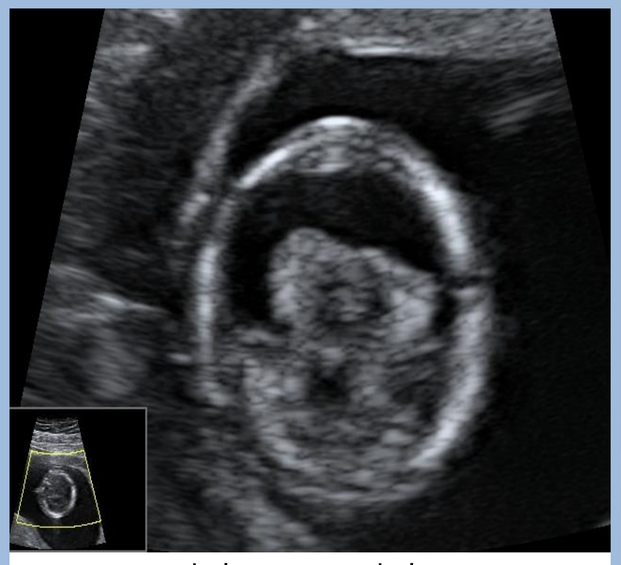
Objective: To describe main pathological ultrasonography findings in fetuses with trisomy 13.

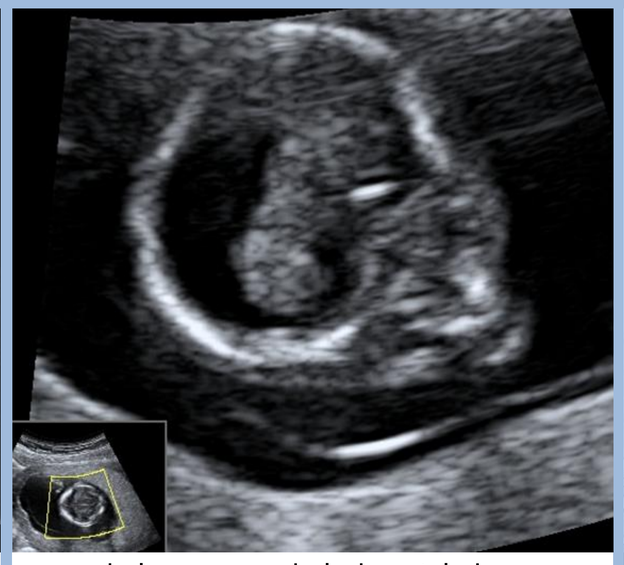
Methods: prospective collection of data during 1st trimester screening examinations in fetuses with trisomy 13 confirmed by CVS or AMC in our department between 1/2011-4/2014. Cut-off point for positivity was the risk 1:100 and higher.

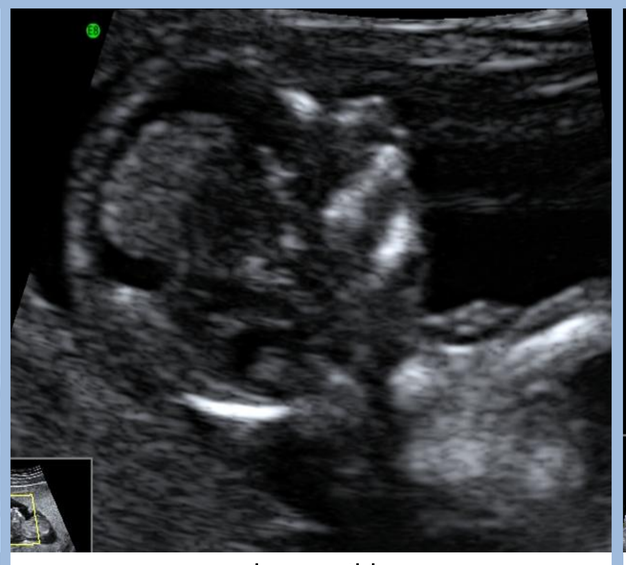
Results: During the study period we performed 5527 first-trimester screening examinations for chromosomal abnormalities. There were 19 pregnancies with positive screening for trisomy 13. All of them underwent invasive diagnostics either by CVS or AMC. Trisomy 13 was confirmed in 6 cases. In each of those 6 cases we described at least 2 abnormal sonographic findings typical for trisomy 13. Altogether we described: hydrops (4x), absent nasal bone (3x), holoprosencephaly (2x), hygroma colli cysticum (2x), gastroschisis (2x), micrognatia (2x), polydactyly (2x), cleft lip and palate (2x), hydronephrosis (1x) and megavesica (1x), hypotelorism (1x).

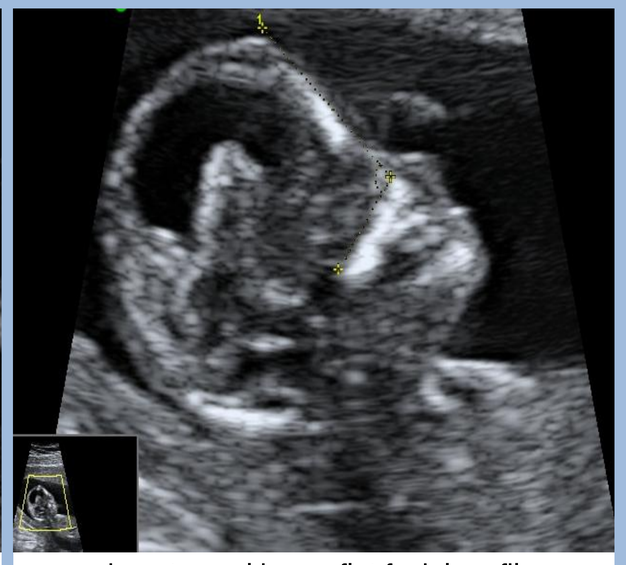

unilateral cleft lip

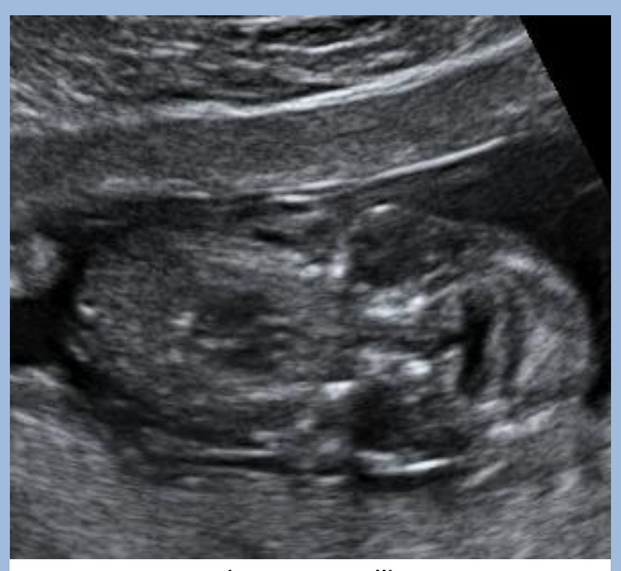

bilateral cleft lip

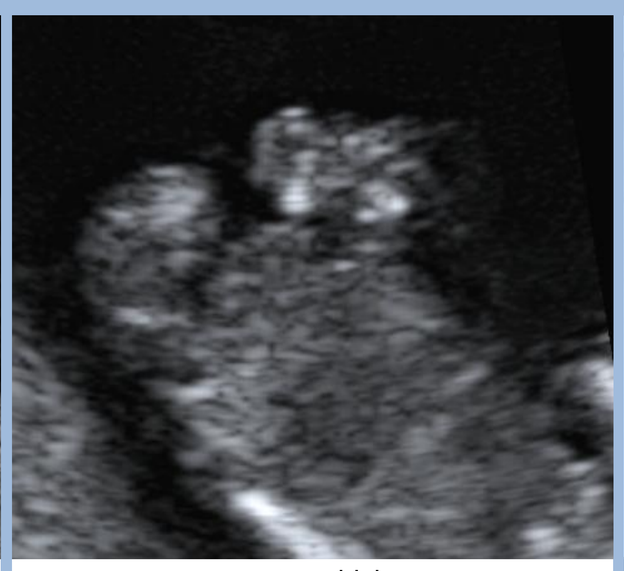

holoprosencephaly

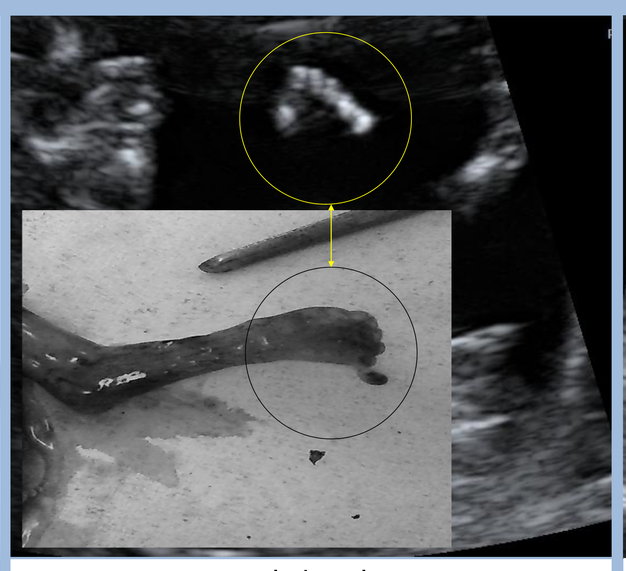

holoprosencephaly, hypotelorism

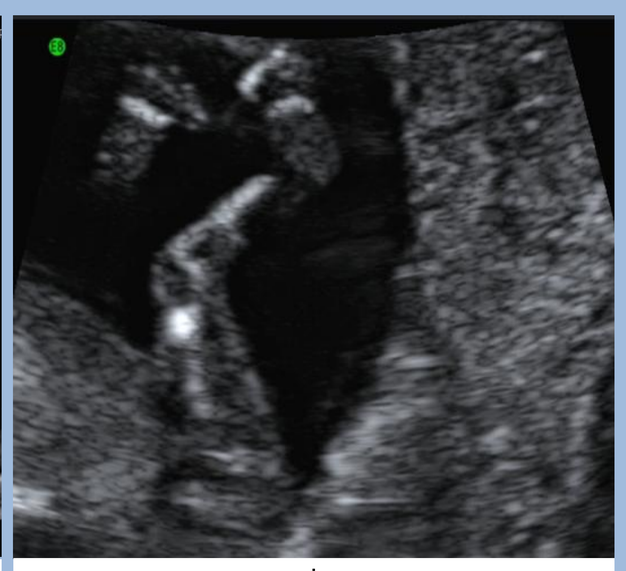

micrognathia


absent nasal bone, flat facial profile


hygroma colli


gastroschisis


polydactyly


pes equinovarus

Conclusion: Trisomy 13 is the third most common aneuploidy with incidence of 1:5000 - 1:20000 newborns. Impaired fetuses are characterized by presence of series of structural abnormalities, typically including holoprosencephaly, hydrocephalus, microcephaly, arhinencephaly, anophthalmia, micrognathia, small or absent nasal bone, cleft lip and palate, hypertelorism, low-set ears, cardiomegaly, dextrocardia, atrial septal defect, ventricular septal defect, severe fetal bradycardia, post-axial polydactyly, flexion of the fingers (different form that of trisomy 18), rocker bottom feet, intrauterine growth retardation, oligo-anhydramnios, polycystic kidneys, hydronephrosis, neural tube defects.
Some of these structural abnormalities are very well detectable by sonography already in the first trimester of pregnancy. Presence of these structural defects should always lead to suspicion of trisomy 13.