A prenatal case of 10q22q23 deletion with heterotaxy syndrome and hypoplastic left heart complex

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Background
Chromosomal microarray analysis has become a routine method in prenatal diagnosis for the cytogenetic detection of copy number variations in fetuses with or without major structural abnormalities. On basis of similar copy number variations in various patients the clinical presentation of these recurrent genomic aberrations are described quite well, whereas chromosomal anomalies, with unpredictable and inconsistent phenotypes, represent a difficult counselling situation for clinicians, especially in the prenatal setting. We present a case with prenatal diagnosis of a 10q22.3q23.2 deletion with clinical features, that not been described so far.

Clinical cases
A 30 year old healthy woman was referred to our department after organ screening with the suspicion of fetal congenital heart defect at 23 weeks of gestation. Ultrasound showed intrauterine growth restriction, a single umbilical artery and unremarkable face profil (Fig 1). Fetal echocardiography revealed a hypoplastic left heart complex with mitral valve stenosis, aortic arch stenosis, subaortic ventricle septum defect, atrial septal defect and tricuspid valve insufficiency. The parents decided to continue the pregnancy. Routine prenatal cytogenetic analysis with Giesma banding through placental puncture revealed a normal female karyotype (46, XX) and thus chromosomal microarray analysis was initiated. The girl was delivered by caesarian section in week 39. On the fourth day of life she underwent Norwood I procedure for univentricular repair. Due to haemodynamic instability an extracorporeal membrane oxygenation was implanted one day after the operation. Four weeks later the girl died because of multiple organ failure and sepsis. Post mortem examination showed heterotaxy syndrome including dextroposition of the stomach, the duodenum and the pancreas and right-sided polysplenia.

Results
Chromosomal microarray analysis revealed a 7.3 Mb deletion on chromosome 10 (10g22.3q23.2). Subsequent chromosomal microarray analyses of both parents showed that the deletion was inherited from the father, who didn’t show any physical symptoms, but mild mental retardation.

Conclusions
We demonstrate the first case of 10q22q23 deletion syndrome leading to a lethal phenotype with hypoplastic left heart complex and heterotaxy syndrome.