The risk of 22q11.2 deletion in fetuses with a right aortic arch and a normal heart

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Objective
To assess the risk of 22q11.2 deletion in fetuses with a prenatal diagnosis of right aortic arch without intracardiac anomalies (RAA-no ICA).

Methods
A retrospective study from 2004 to 2014 of all fetuses with RAA-no ICA diagnosed prenatally in three referral Centres. A detailed sonogram was obtained in each case, including visualization of thymus and of the head and neck vessel to identify the presence of vascular rings (VR). Karyotype and FISH analysis for the diagnosis of 22q11.2 deletion were always offered either prenatally or postnatally. A clinical and echocardiographic examination was performed in live births and a post-mortem examination in case of termination of pregnancy.

Results
22q11.2 deletion was found in 7/82 cases (8.5%, 95% CI 3.8 – 17.3%). The incidence was particularly high when the thymus was small or non-visualized (7/7), and also with abnormal extra-cardiac sonographic findings (3/11) and in the presence of a vascular ring (3/22). The diagnosis of RAA-no ICA was always confirmed after birth.

Conclusion
22q11.2 deletion is present in 8.5% of fetuses with a prenatal diagnosis of RAA-no ICA. The risk is much increased when the thymus is small or absent, and when abnormal extra-cardiac findings or a vascular ring are identified. Conversely, we have never seen 22q11.2 deletion in the presence of a thymus of normal size.