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When is a fetal persistant left superior vena cava truly isolated? A retrospective cohort of 230 cases.

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Basis: Isolated persistent left superior vena cava (PLSVC) is not associated with any cardiac, extra-cardiac or genetic anomaly. The definition of isolated PLSVC varies as some anomalies can be difficult to detect antenatally. Even if not clinically relevant for the patient some of them may be related to a genetic anomaly.
Objectives: To assess the frequency of genetic, cardiac or extra cardiac anomalies in fetuses with isolated versus associated PLSVC.

Methods: Retrospective cohort study including all fetuses diagnosed with a PLSVC between 2010 and 2017. PLSVC was categorised as isolated or associated according to the ante natal diagnosis of associated congenital heart defects, abnormal venous or arterial connections, and/or extracardiac anomalies.

Results: Among 230 fetuses diagnosed with PLSVC, 40 cases (17.4%) were strictly isolated and no syndromic/genetic anomaly or coarctation of the aorta was diagnosed. In the remaining 190 fetuses with PLSVC and associated prenatal ultrasound features, 65 (34%) fetuses had a genetic anomaly: 23 aneuploidies (10 trisomies 21, 7 trisomies 18, 2 trisomies 13, 4 Turner syndrome), 15 pathogenic micro-deletions/duplications (including 3 deletions 22q11.2) and 5 variants of unknown significance. In particular, PLSVC associated with abnormal venous or arterial connections (aberrant sub clavian artery, abnormal ductus venosus) presented a 22.2% rate of genetic anomalies. Small ventricular septal defect, single umbilical artery or hypoplastic aortic isthmus were not associated with higher rate of genetic anomaly. The number of aortic coarctations was low overall (7/230). However, among fetuses without evidence of a major congenital heart defect or extracardiac anomalies, z score of the aortic isthmus diameter inferior to -2 was significantly associated with the postnatal diagnosis of isthmus anomalies (10/19 versus 2/52, $p<0.001$).

Conclusions: Fetuses with strictly isolated PLSVC did not show any genetic or cardiac anomaly. A careful cardiac and morphologic ultrasound examination is paramount as minor variants of the venous or arterial system may increase the index of suspicion for genetic abnormalities and invasive prenatal diagnosis for array-comparative genomic hybridization should then be offered.