Unusual Form of Truncus Arteriosus Associated With 22q11 Deletion

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A 25-year-old woman was referred at 31 weeks’ gestation for prenatal echocardiography because routine obstetrical sonography had detected a cardiac malformation. Pregnancy to that point had been normal. Four-chamber and great-vessel views allowed the diagnosis of truncus arteriosus with an abnormal dilatation of the pulmonary tree (Figure 1 and Movie I). An in situ hybridization study performed on amniocytes culture revealed a de novo 22q11 deletion. The pregnancy was uneventful, and the child was delivered naturally at 37 weeks’ gestation. On postnatal examination, the child carried typical features of DiGeorge syndrome, namely dysmorphism, severe hypocalcemia, and thymic hypoplasia. Echocardiography and angiography confirmed the diagnosis (Figure 2 and Movies II and III). The child died suddenly at two weeks of age, two days before the scheduled surgery. Death resulted from uncontrolled catheter-based sepsis. The parents refused the anatomo-pathological examination.

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