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 dysmorphia, 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.

Figure 1. Truncus arteriosus, type A1, on prenatal echocardiography, equivalent to a four-chamber view. Note the truncus arteriosus above the ventricular septal defect (VSD) and the aneurysmal dilation of the pulmonary tree (PA) as compared with the relatively small aorta (Ao).

Figure 2. Truncal root injection after digital substraction. The truncal root gives rise to a dilated pulmonary trunk and to a normal aortic arch.
Unusual Form of Truncus Arteriosus Associated With 22q11 Deletion
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Circulation. 2002;106:e191
doi: 10.1161/01.CIR.0000046081.44490.8E
Circulation is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75231
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Print ISSN: 0009-7322. Online ISSN: 1524-4539

The online version of this article, along with updated information and services, is located on the World Wide Web at:
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