Sibling recurrence of congenital venous system anomaly with normal karyotype presenting increased nuchal translucency thickness; A case report.

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Introduction
Generally, congenital venous system anomalies including absence of ductus venosus (ADV) and total anomalous pulmonary venous return (TAPVR) are developed in polygenic and multifactorial background. Significance of inheritance factor in those diseases are not fully understood. Reports of sibling or familial recurrence of ADV and TAPVR are quite limited. Hereby, we report a case of recurrent venous system anomalies developed in siblings showing increased nuchal translucency (NT) without aneuploidy.

Case
A primiparous women at age 32 was referred to our hospital for abnormal fetal finding of increased NT (6mm) in the first trimester. Chorionic villus sampling at 14 wks demonstrated normal karyotype. At 23 wks, abnormal dilatated vessel was detected in the dorsal side of the left atrium. (Fig.1,2) In repeated sonographic examinations, ADV and umbilical vein-coronary sinus anastomosis were diagnosed. The mother delivered a male baby (3066g, Aps 8/9) at 39 wks. Except for extended hyperbilirubinemia, the postnatal course of the boy was stable without circulatory problem. The woman was conceived again at age 36. Increased NT (6.5mm) was confirmed as in her previous pregnancy. Normal karyotype was demonstrated in the amniocentesis. At 33 wks, abnormality of pulmonary vein was suspected under ultrasonography (Fig 3). She delivered a male baby (2480g, Aps 8/8) at 38 wks. Infracardiac TAPVR in the neonate was finally diagnosed. Cardiac surgery was conducted on day 15 after birth and the postoperative course was well.

Discussion & Conclusion
Congenital heart disease (CHD) can occur in sibling with elevated risk of 2-5%. Several genetic and environmental factors associated with CHD has been reported. (Table1,2), but the specific factor associated with ADV and TAPVR is not identified. Sibling recurrence in the presented case suggest the possibility that some inheritance factor would affect the development of venous system anomaly.