MP3-2

Persistent Nuchal Translucency And The Fetal Heart

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Background: It has been reported that 6% of nuchal translucency (NT) > 99th centile are persistent into the second trimester. The objective is to describe the outcome of a cohort of cases with persistently elevated nuchal translucency (NT) in the second trimester who underwent fetal echocardiography.

Methods: A retrospective review of cardiac and genetic findings in cases identified with a persistently elevated NT between 2003-2014.

Results: Fifteen cases were identified with a NT >99th centile in the first trimester and persistently elevated to mid-gestation. A third trimester scan was performed in 10 of these cases of whom 8 had persistently elevated NT. In the second trimester, pleural effusions were seen in 6 cases and hydronephrosis in 4. Polyhydramnios was present in 9 cases in the third trimester. Congenital heart disease was diagnosed prenatally in 10/15 cases: pulmonary stenosis (n=5), valve dysplasia (n=2), hypertrophic cardiomyopathy (n=6), aortic stenosis (n=3), ventricular septal defect (n=2), partial atrioventricular septal defect (n=1), aberrant right subclavian artery (n=1). Absence of the ductus venosus was evident in four.
Ventricular hypertrophy, subaortic stenosis and pulmonary stenosis were not present during the first trimester, but manifested in the second or third trimester or in the postnatal period. (Table 1)
On the basis of intention to treat 11/12 (92%) survived to delivery, 9/12 (75%) survived to 28 days and 4/12 (33%) through one year. There was one further death at 14 months. Pulmonary valvotomy was required in three cases and septal myomectomy was performed in two cases.
Eleven out of fifteen cases were genetically confirmed to have Noonan syndrome, one had a normal genetic result and results were not available in three. Mutations identified were PTPN11 (n=6), RAF1 (n=2), RIT1 (n=1) and SOS1 (n=2).

Conclusions:

1. All fetuses with persistently raised NT had evidence of congenital heart disease at birth.
2. There is a high prevalence of Noonan syndrome in cases of persistently elevated NT in the second and third trimester.
3. The outcome in these cases is extremely guarded.

Table 1: Prenatal and postnatal ultrasound findings.

	1St Trimester	2nd Trimester	3rd Trimester	Infancy
Elevated nuchal translucency	15/15	15/15	8/10	-
Pleural effusion	-	6/15	4/10	-
Hydronephrosis	-	4/15	1/10	-
Polyhydramnios	-	-	9/12	-
Valvar stenosis	0/7	3/14	5/8	6/9
Ventricular Hypertrophy	0/7	1/14	3/8	8/9