Disclosure Slide

Financial Disclosure for:
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All authors are employees of Laboratory Corporation of America Holdings ("LabCorp") and may own LabCorp stock.

Outcomes of five pregnancies with increased nuchal translucency, normal chromosomes, negative Noonan mutation analysis, and normal second-trimester anatomy scan

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Introduction

First trimester nuchal translucency measurement is used to identify pregnancies at an increased risk for aneuploidies, isolated heart defects, and certain genetic syndromes (e.g. Noonan syndrome). Although nuchal translucency measurement can offer the patient information that will provide benefits in early decision making, it can lead to difficulties in counseling and increased parental distress. Some studies have noted a risk of approximately 10% for Noonan syndrome in fetuses with increased NT and normal chromosomes. Published reports indicate that if chromosome and structural abnormalities are not present, the majority of pregnancies will have a normal outcome.

Purpose

The purpose of our study was to evaluate postnatal outcomes of fetuses with increased nuchal translucency, normal follow-up diagnostic studies (including karyotype, microarray and Noonan syndrome mutation testing), and normal second-trimester anatomy scans.

Methods

We identified a total of 18 women referred for genetic counseling from 2014 to 2018 due to increased nuchal translucency. These women proceeded to have normal anatomy scans and prenatal diagnosis, including karyotype, microarray and Noonan syndrome mutation panel. Eight of these 18 patients were available to follow-up. Three declined to participate. Long- term data on 5 of these pregnancies is presented.

Results

There were two females, both currently three-years of age, developing normally and reporting no health problems, although one was born with a hemangioma of the left breast. A three-year old male born with ankyloglossia reportedly had problems with nursing and gaining weight. Since repair, by a pediatric dentist, he has had normal growth and development. A nearly two-year old male was followed by neurology due to large head circumference which was determined to be familial and benign. A one-year old boy born at 27 weeks due to PROM reportedly has no current medical concerns. All 5 children in our study do not have serious health issues related to the increased NT identified on fetal sonogram.

Table 1: Summary of prenatal and postnatal clinical information

Case	NT (mm)	CHILD'S AGE (months)	Sex	MEDICAL CONDITIONS	GROWTH AND DEVELOPMENT
1	3.20	36	F	Hemangioma of left breast treated with beta blockers	Normal
2	3.40	36	F	None	Normal
3	3.50	23	M	Evaluated by neurology due to large head circumference. Determined to be familial (benign).	Normal
4	3.60	39	M	Failure to thrive due to ankyloglossia. Repaired by pediatric dentist. Immediate weight gain after surgery.	Normal
5	4.00	17	M	Born preterm at 27 weeks due to PROM.	Normal

Conclusions

One of the goals of this study was to add data to the current research in order to provide patients with more accurate information regarding long term outcomes. Although the numbers in this study are small, this research supports previous studies with similar methods. This body of data indicates that when prenatal diagnostic tests (including chromosomes and Noonan syndrome testing) and anatomy scans are normal, patients may be counseled regarding the chances of a favorable outcome.

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