

## Is karyotype beneficial in prenatal non-lethal anomalies?

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### Abstract

The objective of the study is to evaluate the need for fetal karyotyping after ultrasonographic diagnosis of isolated non lethal anomalies and to stress the importance of prenatal counseling. Sixty expectant mothers were chosen to have detailed fetal anomalies survey including echo were done between 24-28 weeks. The fetuses diagnosed antenately for nonlethal anomalies are subjected to postnatal karyotyping. About twelve cases of isolated nonlethal anomalies were detected. Various anomalies are intra abdominal cyst, VSD, microcephaly, isolated hydrocephalus, cleft lip, cleft palate, talipes, and abnormal wrist position were detected. Out of twelve cases only one case had abnormal Karyotype and it was Trisomy 18. From this study it is concluded, that in isolated Non-Lethal anomalies with otherwise normal morphology, prenatal testing of karyotyping is not beneficial.

**Keywords:** Antenatal diagnosis, Fetal karyotyping, Health management, Non-lethal anomalies.

### Introduction

Prenatal diagnosis has undergone an explosion of growth in the last decade with the advent of improved ultrasound imaging, It is possible to make an intra- uterine diagnosis of fetal anomalies. An ultrasound diagnosis can always be supplemented by fetal echocardiography, when the anomaly is the one that is known to be associated frequently with cardiac disease and fetal karyotyping can be done, using either amniocentesis or fetal blood sampling, when the anomaly is known to be associated with increased frequency of chromosomal abnormalities.

When prenatal diagnosis is made, is often an immutable truth about fetus. Most important in prenatal diagnosis is the management plan regarding timing, and method of delivery after appropriate counseling of the couples taking their wishes into consideration. These advances in prenatal diagnosis have fostered a parallel growth of better techniques of fetal therapy.

### Materials and methods

The study was a prospective study conducted during the period January 2010 to July 2010. Sixty expectant mothers had detailed sonographic fetal anatomic survey including echo done between 24-28 weeks of gestational age with 3.5mhz transducer. Our results were confirmed by the radiologists from the radiology department. The inclusion criteria was, isolated non-lethal anomalies with otherwise normal morphology of fetus. The fetus with lethal anomalies, multiple congenital anomalies, anomalies associated with polyhydramnios oligohydramnios and IUGR were excluded from the study. The mothers who had non-lethal anomalies detected were given repeated counseling and had consultations with the pediatric surgeon as well when needed. All isolated Non lethal anomalies were subjected to postnatal karyotyping just to assess the beneficial role of karyotyping in cases of isolated Non lethal anomalies.

### Results

Out of 60 women, 12 had isolated non-lethal anomalies. 11 cases had normal vaginal delivery with 8.3%; 1 case had elective caesarean section. The baby with isolated hydrocephalus was delivered by elective LSCS to avoid trauma to the brain due to normal vaginal delivery (Conchance & Myles, 1982). Table 1 shows the gestational age at which ultrasonogram was done. At 24 weeks 8 cases, 26 weeks 24 cases and 28 weeks 28 cases had ultrasonogram done. Table 2 shows the list of non-lethal anomalies detected. In the abdominal cavity-1 case of intraperitoneal cyst ,in the cardiac system, one case of isolated VSD in CNS-1 case of isolated hydrocephalus and microcephaly 3 cases, in face-1 cleft lip and 2 cases of Cleft lip and palate and in skeletal system unilateral talipes2 and a case of wrist abnormality. In postnatal karyotyping, 11cases had normal karyotyping while one

Table 1. GA when USG was done

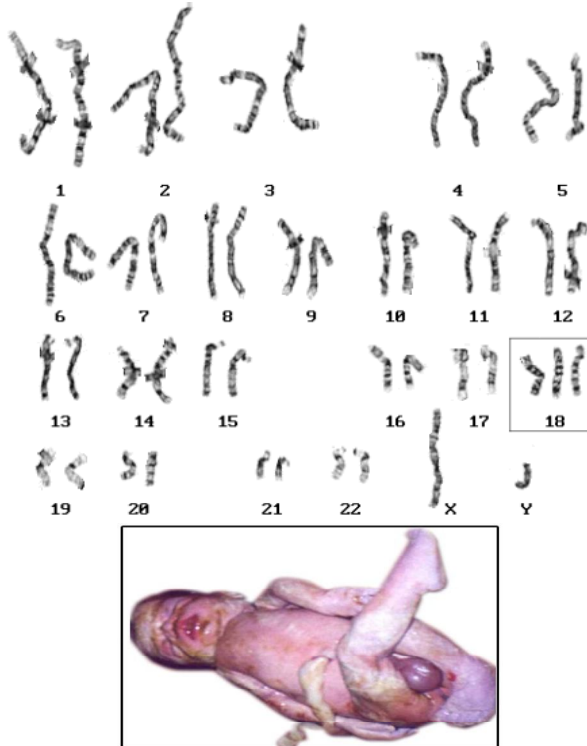
GA	No. of Patients
24 Weeks	8
26 Weeks	24
28 Weeks	28
Total	60

Table 2. Types of non- lethal anomalies

	Detected anomaly	No. of Patients
Abdominal	Intra Peritoneal Cyst	1
Cardic	VSD	1
CNS	Microcephaly	3
	Isolated ydrocephalus	1
Facial	Cleft Lip	1
	Cleft Lip and Palate	2
Skeletal	Talipes	2
	Wrist Abnormality	1
Total		12

case had abnormal karyotyping-Trisomy 18 (Fig.1). Table 3 represents the postnatal follow up. In the intraperitoneal cyst, isolated hydrocephalus, cleft lip and palate were referred to pediatric surgery for subsequent procedures. The case of talipes was advised strapping. The VSD turned out to be a normal heart on postnatal

Fig.1.Wrist abnormality  
(cardiac abnormality missed) - Trisomy 18



scan and the baby with wrist abnormality had pigeon shaped deformity of chest in addition (due to cardiac abnormalities) which was missed antenatally and had intra partum death. The predictive value is given as: Sensitivity -100%, Spcificity-96%, Positive Predictive Value 83.3% and Negative Predictive Value 100%.

**Discussion**

There should be cautious approach when minor malformations are detected and several questions arise after antenatal detection of an anomaly. a). Is the anomaly isolated or associated with other anomalies? b). Is the anomaly compatible with life? c). What are the immediate risks for the fetus? d). What is the likely outcome at birth? e). Is the anomaly correctable after birth? f). What will be the morbidity at birth and later ? and g). Why counseling is important for the parents?

Our study correlates well with wrist abnormality being associated with a high incidence of karyotype abnormality (Paluda *et al.*, 1996). Talipes unless associated with other anomalies does not mandate a karyotyping which goes well with the results (Malone *et al.*, 2000). The isolated cleft lip and small cleft palate are associated with Normal Karyotype according to which can be applied to cases of cleft lip and cleft palate in our study (Berge *et al.*, 2002).

Our decision to deliver the isolated hydrocephalus by elective LSCS at the time of pulmonary maturity and refer to the concerned department for shunt procedures is in

accordance with the findings (Conchance & Myles, 1982).

In our study, repeated consultations reduced the anxiety levels of the parents and were able to take concrete decisions. This is reflected by the results (Aite & Truchhi, 2003). Early diagnosis also allows the parents to organize appropriate neonatal management for sub-lethal and correctable anomalies while termination of early pregnancy is an option for lethal anomalies (Konar, 2002).

Table 3. Post natal follow up

VSD	Normal Heart Postnatal Scan(FP)
Intra- Peritoneal Cyst	Operated-Lympcyst
Isolated	Ref. To Paediatric Surgery
Cleft Lip and Palate	Ref. To Paediatric Surgery
Talipes	Advised Strapping
Wrist Abnormality	Intra Partum death (trisomy 18)FN

**Conclusion**

After prenatal diagnosis of non-lethal anomaly, when detailed sonographic fetal anatomy survey as normal, there is no additional indication for invasive prenatal workup, but certain lethal anomalies should be kept in mind like abnormal wrist position and cystic hygroma, which are often associated with abnormal chromosomes. In these cases, there should be no hesitation for details prenatal workup even though it requires intervention. When the likelihood of outcome is known, it is easy to reassure and counsel the parents, and based on this, it is easier for parents to take concrete decisions.

**References**

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