OBSTETRICS AND GYNECOLOGY

High Incidence of Emergency Cesarean Section Among Fetuses With Unrecognized Chromosomal Abnormalities

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Objective. **To examine the incidence of obstetric complications in fetuses with unrecognized chromosomal anomalies compare with those in which the diagnosis was known before hand.**

Methods. **All cases followed at a private facility in San Juan, PR during the time from January 1993 through February 1997 were evaluated in terms of gestational age and method of diagnosis and eventual pregnancy outcome.**

Results. **There where 9 cases of chromosomal anomalies documented by karyotype analysis among 1377 (0.65%). Among this group, 5 cases were detected by a combination of maternal serum screening, analysis of risk factors and sonography. Among these, 3 cases elected pregnancy termination, one case of trisomy 21was delivered stillborn vaginally at 32 weeks and one case of trisomy 18 delivered vaginally at 29 weeks. Among the 4 cases not recognized prenatally, one case of trisomy 21 was delivered at 27 weeks by classical cesarean section due to malpresentation and 3 cases (2 of trisomy 18 and one trisomy 21) where delivered by emergency transverse cesarean section due to suspected fetal hypoxia.**

Conclusions. **The very high frequency of emergency cesarean section (100%) among fetuses with unrecognized major chromosomal anomalies should make us increase our efforts to obtain at prenatal diagnosis. In all of these cases, a prior diagnosis would have probably avoided a cesarean section and the associated potential maternal morbidity.**

Precis. **The high incidence of emergency cesarean section among fetuses with unrecognized chromosomal anomalies should make prenatal diagnosis of these conditions a primary goal.**

Key words: Cesarean section, Chromosomal anomalies, Down syndrome, Trisomy 21, Trisomy 18, Obtetrical complications

The reported incidence of chromosomal anomalies among most populations varies from 1/526 at age 20 to 1/7 at age 49, an thus, the total incidence in the population will depend on its mean age (1,2). Some controversy exists regarding the prenatal detection of chromosomal anomalies, since the costs of screening methods such as sonography and maternal serum screening are high. If the only indication for detection were to allow for a decision to terminate the pregnancy, in those cases where the patient refuses pregnancy termination, screening would not be performed.

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However, detection of a chromosomal anomaly can help direct pregnancy management beyond decisions such as pregnancy termination. Among fetuses with unrecognized chromosomal anomalies, the incidence of cesarean section has been reported to be 50%, which is higher than the national average of primary cesarean sections (3). Based on these facts, we decided to evaluate the impact of detection of chromosomal anomalies in determining prenatal management.

Material and Methods

Record review of all pregnancies delivered during the period of January 1993 through February 1997 was done in a private patient population. Only pregnancies reaching a gestational age equal to, or greater than 18 weeks were considered. The reason for choosing this gestational age was that at this time maternal serum and sonography screening would have been done in most patients. A total of 1377 cases were evaluated; the presence of documented

chromosomal anomalies, delivery method and complications of delivery where assessed and the results compared between prenatally detected and unrecognized cases.

Results

A total of 1377 deliveries greater than 18 weeks of gestation were managed during this time period. Among these, a total of 9 cases of documented chromosomal anomalies were identified (one trisomy 13, 4 trisomy 18 and 4 trisomy 21). The incidence of major chromosomal anomalies in this population was 1/153 (0.65%) which is similar to reported incidences (1,2).

Five cases were detected prenatally by a combination of risk factor analysis considering age, previous history, maternal serum alfa feto protein screening and sonographic findings of congenital anomalies or markers for aneuploidy. Four cases were not detected prenatally. (Table I)

Among the 5 prenatally detected cases, three (one case each of trisomy 13,18 and 21) terminated their pregnancy prior to 22 weeks of gestation with no resultant maternal morbidity. Two of the prenatally detected cases decided to continue their pregnancy. Of these, one case of trisomy 21 was delivered vaginally at 32 weeks after detection of an intrauterine demise and one case of trisomy 18 was delivered vaginally at 29 weeks after spontaneous preterm labor and died during the neonatal period.

Among cases not detected prenatally, one case of trisomy 21 was delivered by a classical cesarean section due to malpresentation (transverse lie), premature rupture of membranes and preterm labor at 26 weeks of gestation. One case of trisomy 21 was delivered at 38 weeks by a low vertical cesarean section due to malpresentation, oligohydramnios and repetitive heart rate decelerations. Two cases of trisomy 18 were delivered at 36 and 37 weeks by emergency cesarean section due to oligohydramnios and repetitive fetal heart rate decelerations suggestive of fetal hypoxia. Among these cases there where 2 survivors (one trisomy 21 and one trisomy 18) both with significant long-term morbidity. Analysis of these records revealed that 2 of the undetected cases (one case of trisomy 21 and one of 18) had normal sonograms and alfa feto protein screening, a negative medical history and a maternal age less than 35 years. The two other cases (one case of trisomy 21 and 18) showed abnormally low levels of maternal serum alfa feto protein at 16 weeks of gestation. In both cases sonographic evaluation failed to detect any anomalies and the patients decided not to proceed with a genetic amniocentesis mainly, because they did not desire pregnacy termination. (Table I)

Table I. Management and Outcome of Patients with Chromosomal Anomalies

Type of trisomy	Time of detection	Management after diagnosis	Complications of pregnancy	Fetal outcome	Prenatal diagnosis
13	16 wk	TOP at 18 weeks	None	Stillborn	Sono
18	20 wk	TOP at 21 weeks	None	Stillborn	Low AFP
18	17 wk	VD at 29 weeks	None	Died	Sono/low AFP
21	17 wk	TOP at 19 weeks	None	Stillborn	Age
21	20 wk	VD at 32 weeks	Fetal demise, hydramnios	Stillborn	Sono/low AFP
21	PP	Classical CS at 26 weeks	Transverse lie, preterm labor, premature rupture of membranes	Died	Low AFP/refused genetic amniocentesis
21	PP	Low vertical CS at 35 weeks	Breech, oligohydramnios, fetal distress	Survived	None
18	PP	Emergency CS at 36 weeks	Oligohydramnios, fetal distress	Died	None
18	PP	Emergency CS at 37 weeks	Oligohydramnios, fetal distress	Survived	Low AFP refused genetic amniocentesis

*PP=post-partum, TOP=termination of pregnancy, VD=vaginal delivery, CS= Cesarean section, AFP=alfa feto protein, Sono=sonogram

Discussion

Prenatal diagnosis of chromosomal anomalies is a primary goal in prenatal care. Although diagnosis has improved considerably in the last few years, its sensitivity is still far from 100%. Use of a cut off age of 35 as a risk factor for performing a genetic amniocentesis, will only detect approximately 20% of chromosomal anomalies since the majority of these fetuses are born from mothers less than 35 years of age (4). Use of sonography for detection of sonographic "markers" which can suggest the presence of a chromosomal anomaly is limited since only 20 to 40%

of these fetuses will exhibit findings suggestive of aneuploidy between 14 to 22 weeks (4). The use of triple serum screening (alfa feto protein, estriol and human chorionic gonadotropin) can detect approximately 60% of all cases of Down's syndrome and 80% of cases of trisomy 18 (5,6). There are some new modalities which promise an improvement in sensitivity such as first trimester sonographic measurement of nuchal translucency which may detect up to 60% of cases of trisomy 21 and maternal serum screening of metabolites during the first trimester (7). These tests offer promise for the future, however at the expense of requiring testing of at least 5% of the general population.

Some criticism has been made to the intensive efforts directed nowadays towards the diagnosis of chromosomal anomalies. Some investigators argue that since many of these fetuses will die, performing these studies in the population is not cost-effective (8). In addition, many patients refuse performance of these tests due to a belief that they are done with the express purpose of terminating pregnancy in the face of an anomaly. This would be particularly important in dealing with patients who express that they would not terminate a pregnancy in the presence of a congenital anomaly. Abortion is not an option in many latin-american countries making a diagnosis of a chromosomal anomaly less crucial when deciding upon pregnancy management. Some groups criticise population screening methods as a tool to eugenice the population and discriminate against an individual for genetic reasons (9).

Analysis of the above data shows that in the non-diagnosed cases, a prenatal diagnosis would have at least avoided the performance of an emergency cesarean section and therefore prevented the potential maternal morbidity associated to this procedure. Even if pregnancy termination is not considered an option by the patient, prenatal detection can allow for management planning and non-heroic intervention (such as avoiding an emergency cesarean section in a fetus with trisomy 18). In addition, advanced knowledge of the presence of a major chromosomal anomaly can spare the parents the shock of finding out that their newborn baby has a severe or even lethal anomaly.

It may not be possible to improve prenatal detection of chromosomal anomalies significantly beyond the current level. However, the arguments in favor of population screening are valid.

Resumen

El objetivo de este trabajo es examinar la incidencia de complicaciones obstétricas en fetos con anomalías cromosómicas en una población de alto riesgo. Todos los casos que fueron evaluados en una clínica privada en San Juan, Puerto Rico durante el período entre enero 1993 y febrero 1997 se observaron en terminos de edad gestacional, método de diagnóstico y los resultados finales del embarazo. Se encontraron 9 casos de anomalías cromosómicas documentadas por análisis cariotípico entre los 1377 vistos en la clínica (incidencia 0.65%). Entre los 9 casos, 5 fueron detectados por pruebas de sangre en la madre, análisis de factores de riesgo y/o sonografía. Tres de los casos elijieron terminación de embarazo, uno era una trisomía 21 que resultó en un natimuerto tras un parto vaginal a las 32 semanas de gestación, y el quinto caso era una trisomía 18 que terminó en parto vaginal a las 29 semanas. Cuatro de los 9 casos no fueron detectados en el período prenatal; Una trisomía 21 terminó en una cesárea clásica a las 27 semanas debido a la presentación fetal, 3 casos (2 de trisomía 18 y una trisomía 21) necesitaron una cesárea transversa de emergencia debido a la sospecha de hipoxía fetal. La alta incidencia de cesáreas de emergencia entre los fetos con anomalías cromosómicas no detectadas debe motivarnos a redoblar nuestros esfuerzos de detección prenatal. En todos estos casos un diagnóstico temprano pudo haber evitado una cesárea y la morbilidad materna potencial asociada.

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