18th World Congress in Fetal Medicine

# Prevalence and factors associated with congenital malformations in fetal stillbirth

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## Objective

To establish the epidemiological profile of fetuses with congenital malformations and to determine whether the factors associated with more common fetal malformations could guide the implementation of specific preventive measures.

## Methods

A series of retrospective cases of multimalformed fetuses diagnosed by ultrasound and admitted to the obstetric center of Carlos Andrade Marín Hospital, from 2017 to 2018. All fetal losses (miscarriages and stillbirths before 33 weeks) were included.

## Results

Out of the 276 spontaneous fetal losses, 41 fetuses were detected with fetal defects, representing 14.85%. 24.3 % of these mothers had a remarkable personal medical history such as diabetes mellitus, hypertension or hypothyroidism while 31.7% of them had a positive family history of conditions such as diabetes, heart and oncological diseases. 17.07% of those mothers were taking medication other than multivitamins and folic acid and 33% of them had previous history of abortion. Of the patients who had recurrent miscarriages (7 patients), only one had a history of fetal malformation. The first trimester screening test was performed in 16 out of 41 patients with abnormal findings in 5 fetuses. The remaining 25 fetuses were scanned later in the pregnancy due to several reasons such as ignorance of the pregnancy or of the last menstrual period. Out of these 25 cases fetal hydrops was detected in 17 of them (41.46 %) while 13 of them had coexistent major malformations and an abnormal karyotype. 8 cases (47.06%) did not show any other malformations. Structural cardiopathies were identified in 29.2% of the multimalformed fetuses, of which 50% were associated with hydrops. Out of the 16 karyotypes that were investigated 75% resulted to abnormal findings.

## Conclusion

Fetuses with malformations represent 14.85% of fetal losses in women between the ages of 21 and 44 years. The majority of fetuses had an abnormal karyotype, corresponding mostly to women older than 35 years. There were no association between previous remarkable maternal medical history and fetal malformations. Cardiac pathology represented the most frequent defect with half of the cases being associated with fetal hydrops. A multicenter collaborative study is needed to establish local and national prevalence.

| Number of Case | Abnormal Karyotype | Maternal Age | Anatomical Alteration | Gender | Risk Factor |
|---|---|---|---|---|---|
| 1 | 47 XX +21 | 40 | Cystic Hygroma | Male | Maternal Age |
| 4 | 45X | 29 | Hydrops Fetal, Horseshoe kidney, Heart disease | Female | None |
| 5 | 47XY + 21 | 33 | Hydrops Fetal – Heart disease Functional heart disturbances | Male | None |
| 15 | 46, XX/ 45,X | 37 | Giant encephalocele, Eyelid Fusion, Microtia, Heart Disease, Cleft lip-palate | Female | Insulin dependent diabetes mellitus |
| 17 | 47XX + 18 | 34 | Craniofacial anomalies, Heart Disease, Equinovarus foot, Limb spasticity | Female | None |
| 19 | 47XY + 18 | 43 | Craniofacial anomalies, Hydrops Fetal, Heart disease, Umbilical cord 2 arteries | Male | Maternal Age |
| 23 | 45X | 40 | Hydrops- Heart Disease | Femenino | Maternal age |
| 25 | 45X | 23 | Hydrops – No greater malformation | Undeter mined | None |
| 27 | 47,XY+18 | 35 | IUGR, Subaracnoideo Cyst, Cleft lip - palate, Heart Disease, Limb contracture, Syndactyly, Hypoplasia of the penis | Male | None |
| 28 | 47,XX+18 | 34 | Hydrops Heart disease, Mega cisterna magna | Masculino | Ninguno |
| 29 | 47,XY+21 | 42 | Hydrops Fetal, No greater malformations | Male | Maternal Age |
| 30 | 46,XY isocromosoma(9)(q10 q10) | 35 | No Malformation | Male | None |
| 40 | 47,XY,+21 [85%]/46,XY [%15] | 44 | Restriction, Hydrops | Male | Maternal Age |