FCP110

THE INCIDENCE OF CONGENITAL MALFORMATIONS IN GAZI UNIVERSITY HOSPITAL

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Objective: To determine the incidence and types of congenital anomalies in Gazi University Hospital.
Method: The total number of 4261 neonates born in the Department of Obstetrics and Gynecology, Gazi University Faculty of Medicine during 1997-2002 were studied retrospectively. The birth registries of newborns with congenital anomaly were identified. The total incidence, types and combined anomalies were determined.
Results: The overall congenital anomaly incidence was 1.7% and the NTD incidence 0.50% in our population. Anencephaly was the second most common NTDs with the ratio of 37%, following the spina bifida cases. Facial and musculoskeletal system abnormalities were two and third most common malformations. Urogenital system anomalies were fourth most common type of malformation with an incidence 0.1%. Omphalocele incidence population was 3 in 4261 births and gastrochisis was 2 in 4261 births.
Conclusion: The overall congenital anomaly incidence in newborn in our population is 1.7%. The most common malformations were NTD, facial and musculoskeletal system abnormalities. NTDs incidence was 0.50% in Gazi University Hospital.

FCP111

A RARE CASE REPORT: 45,X/47,XXX MOSAICISM IN GTG BANDING AND ADDITIONAL XX/XXXX MOSAICISM IN FISH TECHNIQUE THROUGH AMNIOCENTESIS AND CORDOCENTESIS; A COMPOSITE KARYOTYPE INVOLVING TRISOMY 7 IN CULTURED SKIN FIBROBLASTS

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We report cytogenetic and FISH analyses performed in the amniocentesis (AS) and cordocentesis (CS) material of mosaic sex chromosome and marker chromosome of the fetus in utero combined with trisomy 7 in cultured skin fibroblasts of the same fetus after the termination. A 27 year old, gravida 3, para 1 woman was referred for prenatal diagnosis because of intrauterine growth retardation and abnormal triple test screening indicating an Edwards syndrome risk of 1/83. Amniotic fluid cell culture and cord blood culture were performed synchronously in the 27th week of gestation (21st week of gestation in ultrasound findings). After the GTG banding technique, the fetal karyotypes of the AS were found as 45,X in 14 clones, 47,XXX in 3 clones and 45,X,-16,-X,+mar in 3 clones from 2 separate culture dishes. The fetal karyotypes of the CS were 47,XXX (78%) in 39 clones and 45,X (22%) in 11 clones. Since ultrasound evaluation showed that there had been no uterine growth in the preceding weeks and the delivery of the patient started spontaneously at 32nd week (24th gestational week in sonography), a 900gr ex fetus was given birth. Physical examination of the fetus revealed dismorphic phenotype and external genitalia was evaluated as ambiguous. Autopsy findings were right polycystic kidney and skeletal deformities indicating mesomelic dysplasia. Skin fibroblasts culture was performed and beside the diagnosis of monosomy X, a third cell line, trisomy 7 was detected and karyotype was evaluated as composite; 46~47,X,+7,-X[cp18]. Fluorescence in situ hybridization (FISH) technique was performed to the amniocentesis and cordocentesis materials and a third and a fourth cell line were determined, revealing XX and XXXX karyotypes. It was shown that FISH proved useful in detection of the low frequency cell lines which need analyses of a large number of metaphase spreads by GTG banding.