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# Familial 22q11.2 Deletion in digeorge/ velocardiofacial syndrome: Pregnancy options and management

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DiGeorge/Velocardiofacial syndrome (DGS/VCFS) is caused by deletion of approximately 1.5Mb to 3Mb of DNA in the q11 region of chromosome 22 long arm which can affect about 30-50 genes. This microdeletion can be detected today by implementation of fluorescence in situ hybridization (FISH), which has improved significantly the overall detection rate of this genetic trait. DGS is an autosomal dominant disorder hence, couples in whom either female or male is affected, have 50% risk for an affected child in each pregnancy. Diagnosis of DGS during pregnancy imposes significant dilemmas and emotional stress upon both the parents.

We report here a 21-year-old female in her first pregnancy, delivered a baby boy at term with congenital heart disease with typical findings of tetralogy of fallot, pulmonary atresia and major aorto-pulmonary collaterals. Due to the above clinical findings the baby was referred for genetic FISH testing which revealed the presence of 22q11.2 microdeletion suggestive of DGS. The baby died on postnatal day 40 due to septicaemia in a tertiary care hospital. Genetic FISH testing was performed in parents to establish the transmission of the above microdeletion found in the child and the result turned out to be positive for DGS and the transmission was confirmed to be from mother to child. In her second pregnancy the following year, prenatal invasive FISH testing was done using chorionic villus sample and the presence of 22q11.2 microdeletion was detected. The couple were advised for genetic counselingand after extensive counseling they chose to continue the pregnancy. At 16 weeks, antenatal scan revealed fetal hydrops with bilateral pleural effusion and ascites ending in a miscarriage.

The common approach for prevention of genetic disorders related to chromosomal aberrations is a therapeutic abortion in cases of affected embryos. Definite diagnosis of a chromosomal disorder is made during the pregnancy by invasive procedures by obtaining embryonic cells.

The ability to perform pre-implantation genetic diagnosis (PGD) and to combine genetic tests on single cells with assisted reproductive technology has revolutionized the field of prenatal genetics and now provides a better option for couples in need. Genetic counselling and evaluation of parents with an affected offspring, and the option of pre-implantation genetic diagnosis, donor gametes during assisted reproductive technology and prenatal diagnosis will help the affected couples.