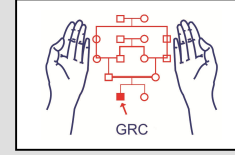


Prenatal Diagnosis of Trisomies

Genetics Resource Centre (GRC)



Prenatal diagnosis of the common trisomies can be done either by the conventional cytogenetic analysis or by PCR amplification of the Short Tandem Repeats (STR). The cytogenetic analysis usually takes longer than the PCR based method. At GRC we have developed sensitive method of detecting the common trisomies (13, 18 & 21) in the CVS DNA. The report is usually available within a few days. The chances of misdiagnosis are around 0.5%.

Who needs the test?

The couples who already have children with Downs syndrome or in whom the screening tests like triple test or ultrasound are suggestive of Downs syndrome are advised to have confirmation by fetal sampling and its testing in the lab. The chances of having a baby with Downs syndrome increase with increasing age of the parents. The couples in age group 35 plus are also advised to have prenatal diagnosis for Down syndrome.

What samples are required?

In addition to the fetal sample we require 2 ml blood in EDTA of the father, mother and the previously affected child (if any). The test can usually be done without the samples of the father and the previously affected child. However, in certain situations the father's sample may also be required for completing the test.

Are there any limitations in the prenatal diagnosis of Trisomies?

The rapid PCR based screening for trisomies can be done in nearly 95% of the couples. However, in the remaining 5% no confirmatory result can be obtained.

Is termination of pregnancy for genetic disorders permissible in Islam?

Most religious scholars in Pakistan, including Molana Mohammad Taqi Usmani, agree that Islam permits termination of pregnancy for a serious genetic disorder in the fetus provided it is done before 120 days (17 weeks) of gestation.

