
mfine
/diagnostics
The
Wait is
Over!
QF-PCR
Fastest Prenatal Diagnostic Test is Here
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What is QF-PCR?

Quantitative Fluorescence Polymerase Chain Reaction (QF-PCR) technique facilitates detection of chromosomal condition in unborn babies using the samples collected through amniocentesis or chronic villus sampling. This is recommended when the non-invasive prenatal screening tests shows an increased risk of chromosomal abnormalities. QF-PCR technology can detect trisomies such as Down syndrome, Edwards syndrome & Patau syndrome, sex chromosome aneuploidies, submicroscopical deletions and duplications.

Conventional diagnostic techniques use fetal cell culture which may require several weeks to obtain the result while QF-PCR does not require cell culture and almost entirely automated to give rapid test results in a time period of 2 days. This technique employs fluorescent bound amplification of specific chromosomal regions, facilitating quantification and the detection of aneuploidy conditions.

QF-PCR Technology Advantage

SPECIFICATION	KARYOTYPE	FISH	QFPCR
AUTOMATION	NO	NO	YES
MCC	NO	NO	YES
TAT	21-30 DAYS	2 DAYS	2 DAYS
SAMPLE TYPE	LIVE CELLS	LIVE CELLS	DNA
CULTURE	YES	YES	NO
SAMPLE REQUIREMENT	HIGH	HIGH	LOW
DIAGNOSTIC	YES	NO**	YES

***ACOG guidelines of 2016 , has considered FISH as a screening tool only for its false positive and false negative results*

Maternal Cell Contamination

One of the risks associated with prenatal testing is maternal cell contamination (MCC), which can occur when a fetal specimen comes into contact with maternal blood or tissue. The risk of MCC is associated with procedures such as chorionic villus sampling, amniocentesis, or extraction of fetal blood from the umbilical cord (cord blood) and in product of conception (POC). If MCC is present, the maternal DNA may mask the results of any genetic testing performed on the fetal DNA thereby increasing the chances of false negative and false positives.

To provide a timely and accurate interpretation, the laboratory and clinicians must be confident that the sample used for analysis is of purely fetal origin.

Contamination of a fetal or cord blood specimen by maternal cells is a potential source of error in diagnostic prenatal testing. Although contaminating maternal blood can be visualized in 1% to 2% of amniotic fluid samples and in up to 38% of pelleted amniocytes following centrifugation,[1] the origin of this blood—fetal or maternal—cannot be reliably assessed. Even low levels of contamination that are below visual detection may negatively impact molecular, biochemical, or cytogenetic results. Highly sensitive molecular testing methods have identified the presence of MCC in 9.1% of direct or cultured fetal cell preparations, 17.8% of which had no visible evidence of maternal blood[2]

TECHNICAL GUIDELINES FOR MCC :[4]

1. MCC testing should be performed on DNA extracted from the same sample or subsample, culture or subculture, that was used for concurrent clinical diagnostic testing.
2. Maternal and prenatal specimens should be tested and analyzed for MCC concurrently within the same analysis to allow for a direct comparison of results.
3. The MCC analysis should use a sufficient number of markers to accurately rule out MCC at the level of sensitivity previously determined by the laboratory during its initial MCC assay validation process.

MFine QF-PCR Advantage

1. First in India to incorporate two sets of markers for increased accuracy, sensitivity and informative result

   - Short tandem repeats (STRs), also known as microsatellites or simple sequence repeats, are short tandemly repeated DNA sequences that involve a repetitive unit. A Short Tandem Repeat (STR) analysis is one of the most useful methods which is used to compare specific loci on DNA from two or more samples. STR analysis measures the exact number of repeating units.

     STR markers are also used in accurate identification and quantification of MCC in prenatal samples thereby providing 100% accurate result

   - Segmental Duplications (SDs) are long DNA sequences (typically defined as being > 1kb in length) that have nearly identical sequences (90-100%) and exist in multiple locations as a result of duplication events.

     Inclusion of SD markers in QFPCR helps in providing informative results in case of consanguineous marriage. In absence of SD markers QFPCR cannot provide informative peaks in consanguineous marriage .

2. Increased Coverage of chromosomal conditions including micro deletions and duplications

MFine Diagnostic QF-PCR Offerings

	QF-PCR EXTENDED	QF-PCR+ MLPA
Coverage	• Trisomies :13, 18, 21, 15,16, 22, Gonosomal Aneuploidies + MCC • DiGeorge Syndrome • Langer Giedion Syndrome • Smith Magenis Syndrome • Miller-Dieker Syndrome • Williams-Beuren Syndrome • Potocki Lupski Syndrome	• Trisomies :13, 18,21, 15,16,22, Gonosomal Aneuploidies + MCC • 20- Common Microdeletions & Duplications

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International Recommendations

SOGC, September 2011
Use of a DNA Method, QF-PCR, in the Prenatal Diagnosis of Fetal Aneuploidies

SOGC recommends QF-PCR as a reliable method to detect trisomies and should replace conventional cytogenetic analysis whenever prenatal testing is performed solely because of an increased risk of aneuploidy in chromosomes 13, 18, 21, X or Y. Both conentional cytogenetics and QF-PCR should be performed in all cases of prenatal diagnosis referred for a fetal ultrasound abnormality or a familial chromosomal rearragement.

Spain, September 2010
Rapid aneuploidy testing versus traditional karyotyping in amniocentesis for certain referral indications

This study suggests that QF-PCR directed to common aneuploidies can be considered as an economically and clinically acceptable prenatal diagnosis policy, offering full karyotype only for specific indications.

India, May 2015
Performance of QF-PCR in targeted prenatal aneuploidy diagnosis: India scenario.

Though QF-PCR is validated in many western countries, there are no studies conducted in India. This research study assessed the utility of QF-PCR as a standalone procedure in Indian clinical set-up. According to the study, the specificity, sensitivity, positive prediction value, and negative prediction values of QF-PCR were 100% while false positive rate and false negative rate were 0% thus, the outcome suggests that this technique can be used as a standalone procedure for targeted rapid aneuploidy diagnosis.

UK, April 2010
QF-PCR as a stand-alone test for prenatal samples: the first 2 year experience in the London region

The reult shows that using QF-PCR as a stand-alone prenatal test for pregnancies without ultrasound abnormalities reduces costs, provides test results and avoids ambiguous and uncertain karyotype reults, thereby reducing parental anxiety and unnecessary terminations.

MFine Diagnostics – MLPA assay for Micro Deletion & Duplication Syndrome

MLPA (Multiplex Ligation-dependent Probe Amplification) is a multiplex PCR method detecting abnormal copy numbers of up to 50 different genomic DNA or RNA sequences, which is able to distinguish sequences differing in only one nucleotide. The inclusion of MLPA in clinical settings therefore significantly increases the detection rate of many genetic disorders. MLPA is a low cost and technically uncomplicated method.

1p36 deletion syndrome	Angelman syndrome
Wolf-Hirschhorn syndrome	Rubinstein-Taybi syndrome
Cri-du-Chat syndrome	Miller-Dieker syndrome
Sotos Syndrome	Lissencephaly-1
Saethre-Chotzen syndrome	Smith-Magenis syndrome
Williams-Beuren Syndrome	Potocki-Lupski syndrome
Williams-Beuren duplication Syndrome	Alagille syndrome
Langer Giedion Syndrome	DiGeorge syndrome
WAGR syndrome	22q11.2 microduplication syndrome
Prader-Willi syndrome	Phalan-McDermid syndrome

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