Neuberg DIAGNOSTICS

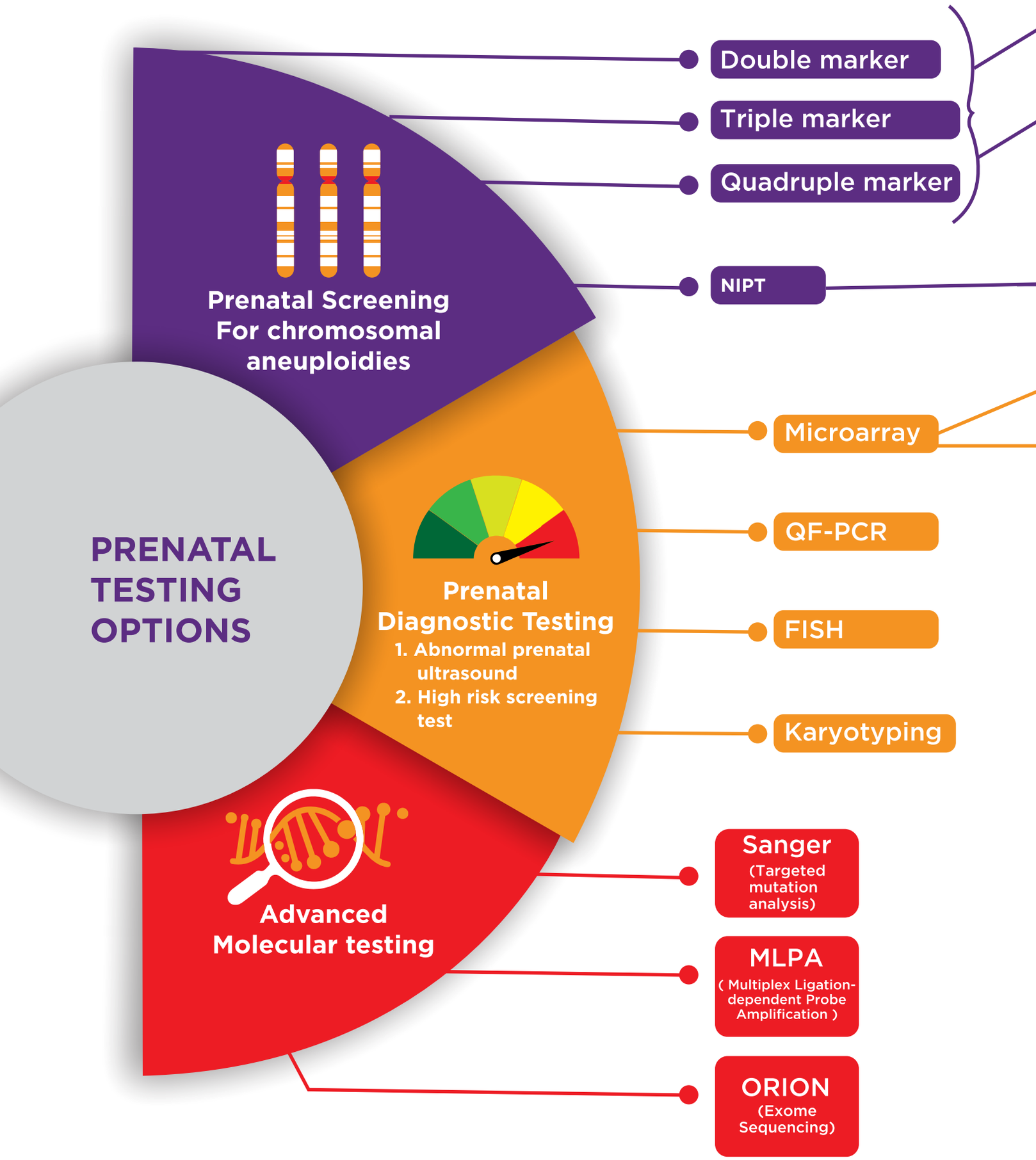
● India ● UAE ● South Africa ● USA

New INSIGHTS

Neuberg DIAGNOSTICS CENTER FOR GENOMIC MEDICINE

Materni - Care

Serial number : 020 Edition : 1. 2022



*Amniotic fluid (AF), chorionic villus sample (CVS), Fluorescent in situ hybridization (FISH)

Low risk → Reduces risk for evaluated aneuploidies

High risk → Further confirmation is commended by AF/ CVS testing

Low risk → Reduces risk for evaluated aneuploidies

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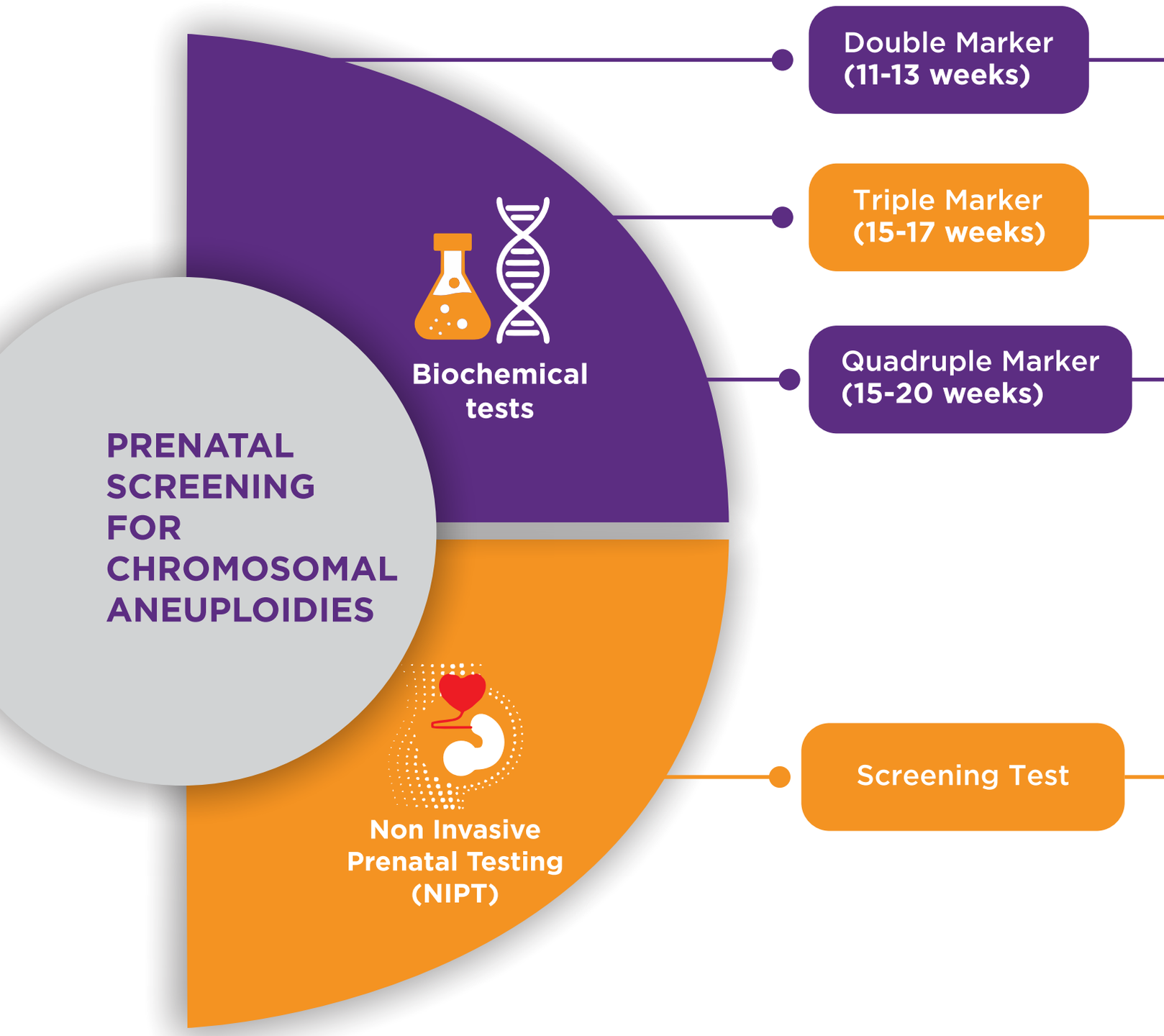
Microarray RapidSure Optima (315K)

Microarray RapidSure Deepdive (750K)

→ Recommended for common microdeletion/ duplication syndromes



Pre - Requisite

- TRF
- NT scan

*Test Requisition Form (TRF), False Positive Rate (FPR), Neural Tube Defects (NTDs)

***ACOG does not recommend prenatal testing for common microdeletions. The sensitivity and specificity for the same is low.**

SCREENING	DETECTION RATE	TAT	LIMITATIONS	SPECIMEN
Trisomy 13/18, 21 and Sex Chromosome aneuploidies	60% FPR ~3% to 5% [1]	48 hours	Screening Test	Maternal Serum
Trisomy 13/18, 21 & NTDs	69% FPR ~3% to 5% [1]	48 hours	Screening Test	Maternal Serum
Trisomy 13/18, 21 & NTDs	80% - 85% FPR ~3% to 5% [1]	48 hours	Screening Test	Maternal Serum

CHROME-Focus:

- Screens for Chromosomal aneuploidies in :
- Chromosome 13 (Patau syndrome)
- Chromosome 18 (Edward's syndrome)
- Chromosome 21 (Down syndrome)
- Sex chromosomal aneuploidies

CHROME-Comprehensive:

- Screens for Chrome focus+ Rare autosomal trisomies

CHROME-Plus:

- Screens for Chrome Comprehensive+ Microdeletions

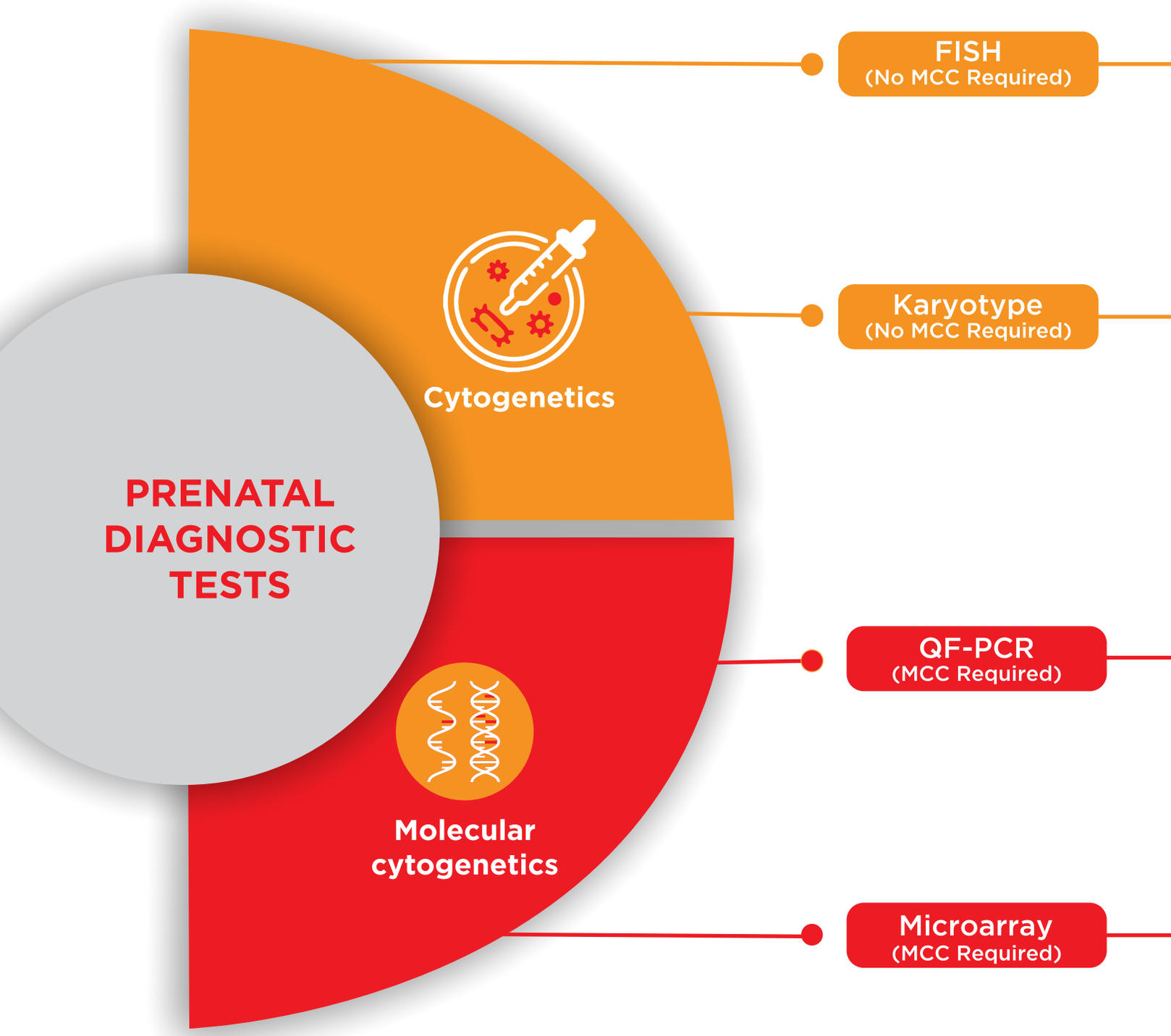
1. DiGeorge(22q11.2)
2. Angelman(15q11.2)
3. Prader-willi(15q11.2)
4. Cri-du-chat(5p),
5. Wolf-Hirschhorn syndrome(4p)
6. 1p36 deletion

DONE AT
After 9 weeks of gestation

TAT
5 to 7 working days

VALIDATED
Singleton/twin pregnancy & donor egg/surrogate

LIMITATIONS
Screening Test



Pre - Requisite
- TRF with clinical details
- Informed consent

MCC
- Form G
- Maternal blood (4ml EDTA)

*Maternal Cell Contamination (MCC), Copy Number Variants (CNVs)

SCREENING	TAT	LIMITATIONS	SPECIMEN
Chromosomal Aneuploidies (13, 18, 21, X & Y)[2]	3 - 5 working days	Cannot detect mosaicism < 10%, limited to the probes used	Blood, AF, CVS
▸ Aneuploidies ▸ Balanced Translocations ▸ Isochromosomes ▸ Ring chromosome[2,3]	15 working days	Small CNVs cannot be detected	Blood, AF
Chromosomal aneuploidies (13, 18, 21, X & Y)	3 - 5 working days	Cannot detect Structural abnormalities & mosaic <30%.	AF, CVS
Chromosomal microdeletions/ duplications/ chromosomal aneuploidies[4] ▸ 315K- 1 MB losses, 2 MB gains & 5 MB LOH / AOH ▸ 750K-200 KB losses, 200 KB gains & 5 MB LOH / AOH	5 - 7 working days	Cannot detect Balanced translocations, mosaic <30%, inversions, small indels and epigenetic alterations	Blood, AF, CVS, Cord

ADVANCED MOLECULAR TESTING

Sanger

Conditions Covered

- Beta thalassemia
- Cystic fibrosis (only Del508 mutation)
- Previously detected pathogenic/ likely pathogenic variants

Multiplex Ligation-dependent Probe Amplification (MLPA)

Conditions Covered

- Atypical hemolytic uremic syndrome (aHUS)
- Duchenne muscular dystrophy (DMD)
- Spinal Muscular Atrophy (SMA)
- BRCA1 &2

ORION

Panels Covered

- Cardiology
- Dermatology
- Endocrinology
- ENT
- Gastrology
- Hematology
- Immunology
- Metabolic
- Nephrology
- Neurology
- Skeletal Dysplasia
- Oncology
- Ophthalmology
- Pulmonology

Sanger analysis

- Specific to targeted genetic variant
- Is only applicable for a specific gene/variant. MCC is required for pre-natal samples.

PRE-REQUISITE
- TRF
- Previous genetic testing report
- Clinical presentation & Family history

TAT
28 working days

SPECIMEN
Blood, AF, CVS

LIMITATIONS:
Cannot capture mosaicism below 15–20%

MLPA

- MLPA is based on PCR principle, useful for the detection of different genetic abnormalities (aneuploidies, gene deletions/duplications, subtelomeric rearrangements, methylation status)[6].
- For disorders where CNVs make up the majority of mutations, MLPA is used as a first-line test.

PRE-REQUISITE
- TRF
- Clinical presentation & Family history

TAT
21 working days

SPECIMEN
Blood, AF, CVS

LIMITATIONS:
Will not detect point mutations, most inversions/ translocations

Next Generation Sequencing

- Most disease-causing variants (85%) are concentrated in the 1–2% of the genome that is protein coding- exons. NGS based exome sequencing involves massive parallel sequencing of upto 20,000 genes.
- Includes multi-exonic copy number variants as well as mitochondrial genome sequencing [2]
- Phenotype specific panel curation possible
- Diagnostic yield of upto 50% [5]

PRE-REQUISITE
- TRF
- Clinical presentation & Family history
- Signed consent form

TAT
28 working days

SPECIMEN
Blood, AF, CVS

LIMITATIONS:
Cannot detect triplet repeat expansions & imprinting disorders

*According to ACMG Guidelines
- Confirmation of the genetic etiology in the proband/affected/index case is necessary.
- If not available, testing of fetal sample along with probands and parents is recommended
- Reproductive decisions based on variants of uncertain significance (VUS) are not recommended.
- Prenatal analysis via Mitochondrial genome sequencing is not available.

Kindly contact the lab before collecting a prenatal sample.

References:

1. The American College of Obstetricians and Gynecologists, Practice Guidelines 2017 (reaffirmed 2020)
2. Lalonde, E., Rentas, S., Lin, F.,et al. Genomic diagnosis for pediatric disorders: revolution and evolution. Frontiers in Pediatrics, 8, 373. (2020) doi: 10.3389/fped.2020.00373
3. Hay, S. B., Sahoo, T., Travis, M. K., et al. ACOG and SMFM guidelines for prenatal diagnosis: Is karyotyping really sufficient?. Prenatal Diagnosis, 38(3), 184-189. (2018) doi.org/10.1002/pd.5212
4. Xia, M., Yang, X., Fu, J., et al. Application of chromosome microarray analysis in prenatal diagnosis. BMC Pregnancy and Childbirth, 20(1), 1-11. (2020) doi.org/10.1186/s12884-020-03368-y
5. Shaffer, L. G., Rosenfeld, J. A., Dabell, M. P, et al. Detection rates of clinically significant genomic alterations by microarray analysis for specific anomalies detected by ultrasound. Prenatal diagnosis, 32(10), 986-995. (2012).doi.org/10.1002/pd.3943
6. Stuppia, L., Antonucci, I., Palka, G., et al . Use of the MLPA assay in the molecular diagnosis of gene copy number alterations in human genetic diseases. International journal of molecular sciences, 13(3), 3245-3276. (2012). doi: 10.3390/ijms13033245
7. Silva, M., de Leeuw, N., Mann, K.,et al. European guidelines for constitutional cytogenomic analysis. European Journal of Human Genetics, 27(1), 1-16.(2019) doi.org/10.1038/s41431-018-0244-x
8. Lalonde, E., Rentas, S., Lin, F.,et al. Genomic diagnosis for pediatric disorders: revolution and evolution. Frontiers in Pediatrics, 8, 373. (2020) doi: 10.3389/fped.2020.00373

In case of any queries kindly contact :
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Reproductive Services

- Maternal Serum Marker Tests
- Non Invasive Prenatal Testing (NIPT)
- Fluorescence in situ hybridization (FISH)
- Karyotyping
- QF-PCR
- Microarray
- Sanger
- Multiplex Ligation-dependent Probe Amplification
- Orion
- Lumos Carrier Screening
- Preimplantation Genetic Testing (PGT)
- Optimal time for Endometrial Receptivity Assay (OPERA)

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