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Microbiological Laboratory

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NABL Accredited Lab

Microdeletion Syndrome

(MDS)

Seen **1** in **2000** births

Why is it **IMPORTANT** to **SCREEN**
for **MDS** ?

MDS may cause severe problems including Severe Learning disabilities (**more severe than Down syndrome**). There is no visual evidence in ultrasound examination. Microdeletions can be de novo mutations with no family history and also not commonly detected with conventional Karyotyping, QF-PCR and standard FISH probes.

List of Detectable Syndromes

1. 15q24 Deletion Syndrome
2. 17q21 Microdeletion
3. 1p36 Deletion Syndrome
4. 22q13/ Phelan McDerimid
5. Cridu Chat Syndrome
6. DiGeorge Syndrome 22q11
7. Langer Giedion Syndrome, 8q
8. Miller - Dieker Syndrome, 17p
9. MECP2 /Xq28 Deletion
10. Prader-Willi/ Angelmen
11. Smith Magenis Syndrome
12. WAGR Syndrome
13. Williams Syndrome
14. Wolf Hirschhorn Syndrome

Test Details

Test Name
MDS

Test Code
90402

Sample
Whole Blood/Amniotic Fluid/ CVS

Methodology
MLPA

TAT
3 Days

DEPRESSING FACT

MICRODELETION SYNDROME is the **SECOND** most common cause of **Congenital Heart Disease** and of **Developmental Delay**

DEPRESSING FACT

Detailed Detection

Detection of aberrant copy number of 50 genomic DNA/RNA sequences in a single, PCR-based reaction.

Accurate Region

MLPA determines the methylation status of imprinted and promotor regions and also detects known point mutations and SNP's

Also Used on Degraded DNA

MLPA can also used on partially degraded DNA, such as DNA extracted from paraffin embedded, formalin treated tissues.



**Genomic Center
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