

|                                                       |                                                  |                                            |                 |                  |
|-------------------------------------------------------|--------------------------------------------------|--------------------------------------------|-----------------|------------------|
| Patient ID<br><b>SA00161024</b>                       | Patient Name<br><b>TESTINGRNV, PADF-ABNORMAL</b> | Birth Date<br><b>1985-06-15</b>            | Sex<br><b>F</b> | Age<br><b>38</b> |
| Order Number<br><b>SA00161024</b>                     | Client Order Number<br><b>SA00161024</b>         | Ordering Physician<br><b>CLIENT,CLIENT</b> | Report Notes    |                  |
| Account Information<br><b>C7028846 DLMP Rochester</b> |                                                  | Collected<br><b>06 Jul 2023 10:45</b>      |                 |                  |

## Prenatal Aneuploidy Detection, FISH

### Result Summary

MCR

**Abnormal**

### Interpretation

MCR

A signal pattern consistent with trisomy 21 was observed, typically associated with a postnatal presentation of Down syndrome.

Chromosome analysis is recommended to distinguish between trisomy 21 and an unbalanced rearrangement to inform recurrence risks. For more information regarding additional testing, call 800-533-1710.

Medical decisions should not be based solely on aneuploidy FISH results per the American College of Medical Genetics and Genomics.

If additional cell cultures have not already been ordered, cultures from this specimen will be discarded 10 days after all cytogenetic test results have been reported. If further testing is desired, contact Mayo Clinic Laboratories at 800-533-1710.

A genetic consultation may be of benefit.

### Result

MCR

| Chromosome | Result        |
|------------|---------------|
| XY         | normal        |
| 13         | normal        |
| 18         | normal        |
| 21         | abnormal(+21) |

### NOMENCLATURE:

nuc ish(DXZ1x1,DYZ3x1,D18Z1x2),(D13S1551x2,D21S339x3)

Of 100 nuclei, 100 had 1 DXZ1 and 1 DYZ3 signal, 100 had 2 D18Z1 signals, 100 had 2 signals for 13q14.3 and 100 had 3 signals for 21q22.

### Reason for Referral

MCR

r/o Down syndrome/Trisomy 21

### Specimen

MCR

Amniotic Fluid

### Method

MCR

| Locus and probes                     | [Strategy;#Nuclei;Class] |
|--------------------------------------|--------------------------|
| XCEN(DXZ1), YCEN(DYZ3), 18CEN(D18Z1) | [COPY#;100;ASR]          |
| 13q14(D13S1551), 21q22(D21S339)      | [COPY#;100;LDT]          |

**Probe strategy includes:** Copy#=region gain and loss.

**Scoring Method:** Manual

### Disclaimer

MCR

Analyte Specific Reagent (ASR) and Laboratory Developed Tests (LDT). This test was developed using an analyte specific reagent. Its performance characteristics were determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration. This FISH test does not rule out other chromosome anomalies (Jalal et al., Mayo Clin Proc 73:132-137, 1998).

### Released By

MCR

Patricia T. Greipp, D.O.

**Received:** 07 Jul 2023 10:48

**Reported:** 07 Jul 2023 11:57

### Performing Site Legend

| Code | Laboratory                                       | Address                                  | Lab Director                | CLIA Certificate |
|------|--------------------------------------------------|------------------------------------------|-----------------------------|------------------|
| MCR  | Mayo Clinic Laboratories - Rochester Main Campus | 200 First Street SW, Rochester, MN 55905 | William G. Morice M.D. Ph.D | 24D0404292       |