Male Infertility Panel

Including: Chromosome Analysis and Y Chromosome Microdeletion (YCMD)
also known as: Molecular Detection of Y Chromosome Microdeletion, Male Factor Infertility, Male Karyotyping, YCMD

PANEL DESCRIPTION:
Our Male Infertility Panel is comprised of both Chromosome Analysis and Y Chromosome Microdeletion (YCMD).
- Each test (Postnatal Chromosome Analysis and Y Chromosome Microdeletion) can be ordered independently if both tests are not desired as the Male Infertility Panel.

TEST DETAILS:
Our Male Infertility Panel includes both Chromosome Analysis and Y Chromosome Microdeletion (YCMD). This panel uses a standard methodology for high resolution chromosome analysis (650 band length or higher) in combination with a multiplex PCR approach. The combination of these tests can detect both structural and numerical chromosomal anomalies and deletions in the Y chromosome including the three azoospermic regions (AZFa, ASFb, and AZFc).

ADVANTAGES:
Influences Clinical Management:
- Men with deletions in AZFa, AZFb, or AZFb+c are poor candidates for sperm retrieval procedures.¹
- Alternatively, men with deletions in AZFc causing azoospermia have a 50% success rate for sperm retrieval.²
- Chromosome Analysis provides the following detection:
  - changes in chromosome number, including aneuploidy and triploidy
  - large scale deletions and duplications
  - balanced and unbalanced structural rearrangements
  - whole genome analysis
- Men who carry structural chromosome rearrangements and their partners may be offered prenatal genetic testing, preimplantation genetic screening, or alternate reproductive strategies.

Determines Risks to Offspring:
- Male offspring of men with AZFc deletions conceived following sperm retrieval will inherit their father’s microdeletion and will have a high risk for infertility.²,³
- Men with structural chromosome abnormalities may have an increased risk for miscarriage and children with chromosome abnormalities or congenital defects.

LIMITATIONS:
- This panel cannot identify all genetic causes of infertility
- Chromosome Analysis will not detect small copy number changes, microdeletions/duplications, single gene mutations, uniparental disomy (UPD), or low level mosaicism
- YCMD is not whole genome analysis
- YCMD will not detect copy number variants (CNVs) in other chromosomes
INDICATIONS FOR TESTING: Genetic testing should be considered for men who exhibit non-obstructive spermatogenic failure (azoospermia or oligospermia). Literature suggests 7% of infertile men have a structural or numerical chromosome anomaly. Specifically, chromosome abnormalities are found in 3-5% of men with oligospermia, 10-19% of men with non-obstructive azoospermia, and less than 1% of men with normal sperm count. Although most often phenotypically normal, an additional 10% of men with azoospermia and severe oligospermia harbor Y chromosome microdeletions. Additional indications for testing include small testes or cryptorchidism and otherwise unexplained male factor infertility.

SPECIMEN COLLECTION & TRANSPORT: Complimentary test kits are available upon request, but are not required.

SAMPLE TYPE and REQUIREMENTS:
- blood:
  - 2-5 ml whole blood in a sodium heparin tube (green top) for Chromosome Analysis
  - 3-5 ml whole blood in an EDTA tube (purple top) for Y Chromosome Microdeletion
  - Send both requirements when ordering the Male Infertility Panel

SHIPPING:
- Coordinate transport for sample to be received in our laboratory within 24-48 hours of collection.
  - LOCAL: Call 402-559-5070 (option 1)
  - OUT OF AREA: Prior to shipment, please fax the completed test request form to 402-559-7248, including the FedEx airbill tracking number.
    - Saturday delivery MUST be checked when sending FedEx on Friday.
    - Please include Internal Billing Reference # 3155070600 on the FedEx airbill.
    - Ship To: Human Genetics Laboratory – Zip 5440
      UNMC Shipping & Receiving Dock
      601 S. Saddle Creek Road
      Omaha, NE 68106

FORMS FOR TESTING: The following forms can be downloaded via our website.
- Required: Male Infertility Test Request Form
- Optional: Informed Consent for Genetic Testing

POTENTIAL TEST RESULTS:

NORMAL:
- A normal (or negative) result indicates that neither a chromosome abnormality nor a Y chromosome microdeletion is a likely cause of the patient’s infertility.
  - 46,XY (male)

ABNORMAL:
- An abnormal (or deleted) result indicates that studies detected a possible cause for the patient’s infertility.

TURNAROUND TIMES: Results are typically available in 1-2 weeks.
BILLING: Our laboratory offers patient/self-pay, insurance (including Medicare/Medicaid), and client/institution billing options. Verifying coverage requirements or obtaining preauthorization PRIOR TO OR AT THE TIME OF SPECIMEN COLLECTION is often necessary. We provide preauthorization services upon request by calling 402-559-5070 (option 3); the following form is helpful for obtaining the information required by insurance providers and can be downloaded via our website.

- Request for Preauthorization

In some circumstances, a test may be warranted even though insurance coverage is denied or not guaranteed. For these situations, we request the following form be signed by the patient and submitted with the sample. This helps inform patients of their potential financial responsibility, should the costs of genetic testing not be paid by their insurance provider.

- Advanced Beneficiary Notice of Noncoverage (ABN) – required when billing Medicare

CPT CODES:
Though some may not be billed, we recommend submitting all listed codes for preauthorization.

- Chromosome Analysis: 88230(2), 88262, 88280
- YCMD: 81403

When applicable, the following codes may be used in place of or in addition to standard package codes: 88261, 88285(3), 88289.

PRICING: For current costs contact the laboratory billing staff at 402-559-5070 (option 3).

REFERENCES:
4. According to the Male Infertility Best Practice Policy Committee of the American Urological Association and the Practice Committee of the American Society for Reproductive Medicine.


updated 4/2018