FISH
also known as: Fluorescence in situ hybridization (FISH), Constitutional FISH, trisomy panel or FISH for aneuploidy, FISH for microdeletions/microduplications, del/dup FISH

TEST DESCRIPTION:
• **Aneuploidy FISH** is used to detect trisomy 13, 18, 21, and aneuploidies of X & Y. This test is not designed to detect structural chromosome abnormalities and inconclusive results may occur if mosaicism is present.
• **RAPID Aneuploidy FISH** will be performed when samples are received on newborns 3 months of age or younger, enabling results to be available within 24-48 hours of specimen receipt.
• **Constitutional (Targeted) FISH** is used to analyze a particular region of interest within a single chromosome. Alternate testing may be appropriate, as many of the microdeletions/microduplication conditions detected by targeted FISH are also identified using the High Density SNP Microarray.

TEST DETAILS and COMPLEMENTARY TESTING:
• **Postnatal Chromosome Analysis** for confirmation and clarification of trisomy, translocation, or mosaicism of 13, 18, 21 or sex chromosome differences is required in conjunction with Rapid Aneuploidy FISH.
• **Postnatal Chromosome Analysis** is recommended in conjunction with any Constitutional (Targeted) FISH.
• If **single gene testing** is considered for a familial genetic condition or abnormal ultrasound findings, contact a laboratory genetic counselor for coordination of testing prior to sending the sample.

ADVANTAGES:
• Detects aneuploidy, triploidy, structural rearrangements (balanced and unbalanced), and large-scale deletions and duplications
• Rapid results available for newborns
• Testing available on fresh or fixed tissues

LIMITATIONS:
• Targeted assay that provides information about the region(s) of interest only
• Detection dependent on the location and size of the aberration relative to the probe

INDICATIONS for ANEUPLOIDY FISH:
Postnatal Chromosome Analysis is required when ordering this test.
• Physical exam suggestive of Down syndrome, trisomy 13, 28, or sex chromosome aneuploidy (X,Y)
• **RAPID Aneuploidy FISH** will be performed when samples are received on newborns 3 months of age or younger, enabling results to be available within 24-48 hours.

INDICATIONS for CONSTITUTIONAL (TARGETED) FISH:
Postnatal Chromosome Analysis is recommended when ordering this test.
• Multiple congenital abnormalities with or without intellectual disability that suggest a specific microdeletion or microduplication
• Family history of a prior child with a previously identified microdeletion/microduplication or familial chromosome abnormality
AVAILABLE FISH PROBES:

All probes in the FISH test catalog below are available for assessment of chromosomal abnormalities based on family history or suspicious/abnormal cytogenetic findings. Please contact our laboratory for case-specific probe availability.

* designates custom probe

- *Charcot-Marie-Tooth / HNPP [17p12]
- Cri-du-Chat [5p15.2]
- DiGeorge / 22q11.2 Deletion GeneReviews®  http://www.ncbi.nlm.nih.gov/books/NBK1523/
- Kallmann [Xp22.3]
- *Langer-Giedion [8q24]
- Miller Dieker [17p13.3]
- *Pallister-Killian [12p]
- Prader-Willi / Angelman [15q11.2]
- SHOX [Xp22.3/Yp11.3]
- Steroid Sulfatase (STS) [Xp22.3]
- SRY [Yp11.3]
- *Waardenburg Type III [2q36-37]
- XIST, X inactivation site [Xq13]
- Centromere enumeration
- Subtelomere analysis for each chromosome
- Whole chromosome Paint Probes

SPECIMEN COLLECTION & TRANSPORT:

Complimentary test kits are available upon request, but are not required.

SAMPLE TYPE and REQUIREMENTS:

- blood, > 3 months of age: 3-5 ml whole blood in a sodium heparin tube (green top)
- blood, newborn: 1-3 ml whole blood in a sodium heparin tube (green top)
- buccal mucosa swab: 5 swabs
- tissue / skin: ≥ 5 mm³ tissue from biopsy or skin punch (transport at room temperature in tissue culture media)

SHIPPING:

- Maintain and ship samples at room temperature.
- 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
REQUIRED FORM: The following form can be downloaded via our website.
• Postnatal Test Request Form

OPTIONAL FORM:
• Informed Consent for Genetic Testing

POTENTIAL TEST RESULTS:

NORMAL RESULTS:
• A normal result indicates no clinically-significant chromosome abnormalities were identified.
• 46, XX (female)
• 46, XY (male)

ABNORMAL RESULTS:
• An abnormal result indicates that a chromosome abnormality was identified that likely provides an explanation for the clinical indications.
  o Parental testing (Postnatal Chromosome Analysis on maternal and/or paternal blood) may be recommended in order to clarify whether the result is de novo or familial for the purpose of recurrence risk calculation.
• Deletion: Missing pieces of chromosomes and/or genetic material. Some may be small and difficult to detect.
• Duplication: Extra genetic material which may be present on any chromosome, such as the presence of two horizontal bands at a specific location instead of one.
• Monosomy: The absence of one whole chromosome. This includes Turner syndrome, a female with a single X chromosome (45,X) instead of 46,XX. Most other monosomies are not compatible with life.
• Rearrangement: Genetic material is present on a chromosome but not in its usual location.
• Translocation: Translocations result when pieces of chromosomes break off and reattach to another chromosome. If it is a one-to-one switch and all of the genetic material is present (but in the wrong place), it is said to be a balanced translocation. If it is not, then it is called an unbalanced translocation.
• Trisomy: The presence of an extra chromosome; a third instead of a pair. These include Trisomy 21 (Down syndrome), Trisomy 13 (Patau syndrome), Trisomy 18 (Edwards syndrome), and Klinefelter syndrome, a male with an extra X chromosome (47,XXY) instead of 46,XY, and other less common trisomies.
• In some circumstances, a result will be unavailable.

TURN-AROUND-TIMES: Results are typically available in 24-48 hours for Rapid Aneuploidy FISH performed on newborns 3 months of age or younger; Results for routine Constitutional (Targeted) FISH are typically available in 7 days when performed on blood and buccal swab samples; Results are typically available in 7-10 days when performed on tissue/skin.

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 Pre-Authorization for Genetic Testing (Postnatal Diagnoses on Peripheral Blood)

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: Due to the complexity of FISH code application, if you have questions please contact a member of our billing team prior to sending a sample.

- blood
- buccal mucosa swab
  - Aneuploidy for 13, 18, 21, X, and Y: 88230, 88271(x5), 88275(x2)
  - Aneuploidy for 13, 18, 21, X, Y, and 22q11.2 analysis: 88230, 88271(x7), 88275(x3)
  - Constitutional (Targeted) FISH: 88230, 88271(x1-3), 88275
    - For each additional chromosomal target: codes above, plus 88271(x1-2), 88275(x0-1)
- tissue / skin: 88271(x2), 88275

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

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