



## EXCEPTION HANDLING FOR ARUP GENETIC TESTING

If you would like to request training or additional information regarding ARUP's process for exception handling of genetic tests, please contact Integrated Oncology and Genetics Services at (800) 242-2787, ext. 3301.

Because genetic testing is generally more complex than other laboratory testing, it poses unique challenges for the clinical laboratory. To handle these challenges efficiently, ARUP has established Integrated Oncology and Genetics Services (IOGS), an exception handling group that deals exclusively with genetic tests and works closely with ARUP's 12 laboratory-based genetic counselors to support clients' genetic testing needs.

ARUP Genetics Services, which includes IOGS and ARUP genetic counselors, offers not only testing services but also brings value to our clients and ordering physicians by ensuring that physicians order correct laboratory tests for their patients.

Genetic testing often requires knowledge of a patient's clinical and family history to determine the most appropriate test and provide the most comprehensive results. Complex genetic test orders are reviewed upon arrival at ARUP to determine if all necessary patient information has been provided with the specimen. This is done to:

- Ensure that the laboratory has the clinical and family history information necessary to provide the most comprehensive and accurate results.
- Assess whether the test order is the most cost-effective option.
- Determine whether the test order is the best option for the clinical situation.

ARUP instituted this review process based on years' experience handling complex genetic cases. These efforts have reduced unnecessary testing and assisted with proper patient diagnosis.

The following three examples illustrate the impact clinical and family history information has on patient care:

As the cases above demonstrate, having proper documentation of the patient's clinical and family history is critical in providing the accurate results. If this information is not included, ARUP's IOGS and genetic counselors will contact the client to request the missing information. This allows ARUP's clients to provide the highest quality care to their patients.

- Hereditary hemorrhagic telangiectasia (HHT) sequencing was ordered when the physician intended to order the hemochromatosis mutation panel.

**Impact:** Because the ordered test was for a different disease, a negative result would have been misleading.

- Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants, Fetal, was ordered for prenatal diagnosis in a fetus of parents with known mutations; parental mutations were not provided with the fetal specimen.

**Impact:** Had the test been run as ordered, the result would have been negative, but the fetus would still have been at 25 percent risk of being affected with CF, since parental mutations are not tested as part of the 165 mutation panel.

- *MSH6* sequencing and deletion/duplication were ordered on a patient with no clinical information provided. Upon inquiry, the lab was told that there is a known Lynch syndrome mutation in the family, and documentation of the mutation was requested. The lab report indicated that the familial mutation was in the *MSH2* gene, not the *MSH6* gene.

**Impact:** Had the test been run as ordered, the patient would have received a negative result and false reassurance of not having the familial Lynch syndrome mutation.

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