



Value of Genetic Counselors in the Laboratory

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ARUP LABORATORIES

ARUP Laboratories is a national clinical and anatomic pathology reference laboratory and a nonprofit corporation affiliated with the University of Utah and its Department of Pathology. With nearly 3,000 employees, ARUP offers in excess of 3,000 tests and test combinations, ranging from routine screening tests to esoteric molecular and genetic assays. Rather than competing with its clients for physician office business, ARUP chooses instead to support clients' existing test menus by offering complex and unique tests, with accompanying consultative support, to enhance their abilities to provide local and regional laboratory services. ARUP's clients include many of the nation's university teaching hospitals and children's hospitals, as well as multihospital groups, major commercial laboratories, group purchasing organizations, military and other government facilities, and major clinics. In addition, ARUP is a worldwide leader in innovative laboratory research and development, led by the efforts of the ARUP Institute for Clinical and Experimental Pathology®.

INTRODUCTION

Genetic counselors (GCs) employed by diagnostic laboratories may write medical papers, coordinate research, create and maintain genetic databases, educate clients and health care providers, and review test orders. Of these duties, the one that most directly benefits patients, medical institutions, and insurers is the rigorous reviewing of genetic test orders. GCs at ARUP Laboratories, a national reference laboratory, collectively save ordering institutions more than \$30,000 per month by modifying test orders to improve utilization.

Seven GCs at ARUP Laboratories performed a review of all genetic test modifications over an 11-month period, reviewing clinical information that accompanied test orders for complex genetic tests (i.e., sequencing, large duplication/deletion analysis, or array-based technologies) before testing was performed. The GCs considered the clinical utility and cost-effectiveness of the ordered tests and contacted the ordering institution and/or health care provider to collect additional clinical information, confirm testing, or suggest alternative testing based on the provided clinical information or family history.

The GCs identified and cancelled or changed inappropriately ordered genetic tests for an average cost savings of \$36,500 per month, representing approximately 30 percent of all complex genetic tests ordered. Among frequently misordered tests were requests for full-gene sequencing when a familial mutation was known or when a screening panel would have been more appropriate (e.g., cystic fibrosis testing in expectant individuals with no family history).

Erroneously ordered genetic testing delays medical decision-making and increases diagnostic costs. In 2008, U.S. health care spending was the highest of all industrialized countries, about \$7,681 per resident, and accounted for 16.2 percent of the nation's gross domestic product (GDP). Reducing the growth in health care costs is thus a priority.

METHODS

Seven ARUP GCs reviewed daily test orders for complex biochemical, cytogenetic, and molecular genetics tests between February 2010 and December 2010. Data regarding genetic test changes prompted by the review was extracted from ARUP's laboratory information system using software designed to identify and tag special characters included in a test-change request. Each case was reviewed to ensure that the reason for the test change was correctly categorized.

Complex Biochemical Genetic Orders Reviewed

Biotinidase

Galactosemia

Hydroxyproline

Mucopolysaccharide

Molecular Orders Reviewed

Alpha Thalassemia
Alport Syndrome, X-Linked
Beta Globin
Biotinidase
Cerebral Cavernous Malformation
Cystic Fibrosis
Familial Mediterranean Fever
Familial Mutation Targeted Sequencing
Freeman Sheldon Syndrome
Galactosemia
Glaucoma, Primary Congenital
Hearing Loss
Hemophilia A
Hemophilia B
Hereditary Hemorrhagic Telangiectasia
HNPCC/Lynch Syndrome
Juvenile Polyposis
Legius Syndrome
LMNA-Related Disorders
Loeys Dietz Syndrome
Medium Chain Acyl-CoA Dehydrogenase
Multiple Endocrine Neoplasia, Type 2
Neurofibromatosis, Type I
Noonan Syndrome
Pancreatitis
Primary Carnitine Deficiency
PTEN-Related Disorders
Pulmonary Arterial Hypertension
RASAI-Related Disorders
Rett Syndrome
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency
Von Hippel-Lindau

Cytogenetics Orders Reviewed

Chromosome Analysis
Genomic Microarray

RESULTS

GCs assisted in an average of 107 test modifications per month, for an average monthly cost-savings of \$36,451 from cancellation of misordered tests (table 1).

table 1

Month in 2010	Cost Savings	Number of Tests Changed
February	\$23,347	72
March	24,330	74
April	48,235	119
May	23,607	105
June	35,779	98
July	31,925	99
August	38,432	110
September	43,207	117
October	38,656	122
November	56,510	149
December	31,928	110
Total	\$402,357	1188
Average per month	\$36,451	107

Test order modification were often the result of misordered tests; the specific types of misorders are shown in table 2.

table 2

Percent of all Cancellations	Misorder Category
37%	Ordered incorrect test; added appropriate test
31%	Ordered incorrect test; no additional testing ordered
11%	Ordered gene sequencing; added targeted panel
10%	Ordered gene sequencing; added targeted test for familial mutation
9%	Test previously performed/duplicate order
2%	Sample was compromised or insufficient
1%	Other reason

Most commonly, the wrong test was ordered, which resulted in test cancellation and ordering of a replacement test. This occurred in over one third of misorders. An example of this includes ordering hereditary hemorrhagic telangiectasia sequencing when the provider intended to order the hemochromatosis mutation panel.

The second most frequent type of misorder resulted in test cancellation with no additional test ordered (approximately 31 percent of orders). In 11 percent of order errors, a sequencing test was ordered when a mutation panel was more appropriate.

Finally, in about 10 percent of misorders, full-gene sequencing was ordered when the familial mutation was known.

Table 3 shows the five most frequently misordered tests by volume.

table 3

Ranking by Volume Cancelled	Test Name	Percent of Specific Test Cancelled
1	Alpha Globin Sequencing	64%
2	<i>NF1</i> Deletion/Duplication	80%
3	Galactose-1-Phosphate Uridyltransferase	16%
4	Targeted Sequencing for Familial Mutation	13%
5	Cystic Fibrosis Panel with Reflex to Sequencing	17%

Our data demonstrate that alpha globin sequencing is often ordered when the provider wants alpha globin deletion testing.

- Neurofibromatosis type I (*NF1*) deletion/duplication testing was the second most commonly misordered test as practitioners usually desired *NF1* sequencing, which is not currently a test performed at our laboratory.
- Galactose-1-phosphate uridyltransferase was the third most commonly misordered test, as most practitioners intended to order galactomannan.
- Familial mutation targeted sequencing was incorrectly ordered by practitioners when there is a familial disease, but no one in the family has previously had genetic testing and the familial mutation is unknown.
- Cystic fibrosis (CF) panel with reflex to sequencing was often erroneously ordered when the practitioner only wanted a CF panel for a routine carrier screen.

SUMMARY

The misordering of genetic tests is a common occurrence in diagnostic laboratories. ARUP Laboratories' genetic test cancellations and additions have enabled us to understand which tests are most commonly misordered and to implement changes in our procedures.

As an example, we are adding a statement to the ARUP Laboratory Test Directory that clients should contact an ARUP GC before ordering alpha globin sequencing. We hope this will significantly reduce the number of inappropriate orders for this test. We have also decided to remove *NF1* deletion/duplication testing from the ARUP Online Test Directory until we can offer *NF1* sequencing. We plan to continue to monitor the most frequently misordered tests and institute changes intended to significantly reduce these errors. ARUP Laboratories' GCs saved hospitals and patients nearly one half million dollars in inappropriate genetic test orders in 2010.

Given that genetic testing will continue to be misordered, it is critical to have personnel who understand the complexities of genetic testing in the laboratory to evaluate genetic test orders. GCs are well qualified for this role, and, by reviewing genetic test orders, can help reduce unnecessary costs to hospitals, laboratories, and patients, thereby improving patient care and reducing the waste of health care dollars. Anything less is unacceptable in this day of upward-spiraling health care costs.

REFERENCES

1. Scacheri C, et al. Molecular testing: improving patient care through partnering with laboratory genetic counselors. *Genet Med* 2008;10(5):337-42.
2. Centers for Medicare and Medicaid Services, Office of the Actuary, National Health Statistics Group, National Health Care Expenditures Data. January 2010.
3. Mayo Clinic. Common Test-Ordering Errors, Part 3: Misordered Tests. Known mutations, molecular vs. nonmolecular test for given disease, chromosome analysis. www.mayomedicallaboratories.com/articles/hottopics/2010-06b-orderingtests-pt3.html (accessed March 15, 2011).

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