Project Title: Increasing the Efficacy of Molecular Diagnostic Testing for Patient Care and Research

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Topic Category:  Clinical

Background, Significance of Project: Molecular diagnostic testing, including exome and genome sequencing, is rapidly increasing our ability to diagnose disease. Accuracy, sensitivity, and scope of disease amenable to diagnosis have been improved. However, molecular testing can produce large-volume data of varying quality, so standardization of the primary data and analysis is warranted. Also, integration of results into the medical record is a challenge that has not yet been solved. Patients with rare diseases are seen at the Clinical Center, and investigators in the 27 research institutes and centers develop “First in Human” clinical trials based upon their research. The research and unique patient population provide an enormous opportunity for medical discovery. As part of these studies, the NIH intramural investigators order thousands of exome/genome sequences each year, often independently of the hospital laboratory. Clinical benefits and research findings may be missed with the decentralized results and with varying quality and analysis tools.

Purpose/Objectives: The purpose of this project is to increase the efficacy of molecular diagnostic testing on campus. Objectives are to (1) assess the volume and type of tests needed, (2) to develop a plan for integrating more results into the patient medical record, and (3) to improve data quality that will enable consistent, cross-institute data analysis.

Methods/Approach/Evaluation Strategy: Interviews were conducted with stakeholders at NIH and with premier academic sequencing laboratories across the country. Testing volume was estimated from billing and contract records, and a Needs Assessment Survey was conducted. Literature was reviewed for best practices, and information was obtained from commercial laboratories to identify the breadth of testing options.

Outcomes/Results: Stakeholders indicated that the use of multiple different laboratories for testing is not always ideal for patient care or for research. Many requested a centralized service within the hospital to barcode samples, and to either perform testing in-house or to coordinate the testing sent to reference laboratories, including data integration into the patient record; however, the financial separation of the institutes creates a challenge to implementing a centralized service.
Increasing the Efficacy of Molecular Diagnostic Testing for Patient Care and Research

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BACKGROUND
Molecular diagnostic testing is rapidly increasing our ability to diagnose disease. The research mission and unique patient population at the NIH provide an enormous opportunity for medical discovery. Although intramural investigators order many sequencing tests each year, clinical benefits and research findings may be missed due to decentralized processing and varying analysis tools.

OBJECTIVES
Standardize the processes for molecular diagnostic testing across the NIH campus.
Aims:
(1) Assess the volume and type of tests needed.
(2) Integrate molecular results into patient medical record.
(3) Enable consistent, cross-institute data analysis.

METHODS
• Interviews conducted with stakeholders.
• Needs Assessment Survey conducted with NIH investigators.
• Literature reviewed for best practices.
• Information obtained from commercial laboratories.

RESULTS
• Consensus from stakeholders was to improve testing coordination.
• Most sequencing results are not routinely integrated into the medical record.
• New NIH database, the Collaborative Research Exchange (CREx), could be expanded to capture testing type, volume, and vendor.
• Lack of a geneticist coordinating testing within the CC.
• Potential cost savings by combining contracts.
• Secondary findings are not assessed in many cases.

Many different reference laboratories used by institute investigators independently of the Clinical Center.

DISCUSSION
Proposal to Leadership:
(1) Hire an expert, such as a Chief Genomic Officer, within the Clinical Center.
(2) Create an advisory group of experts on campus to guide testing practices.
(3) Expand capabilities of existing CREx database to better capture testing on campus.

FUTURE IMPACT
Benefits:
-Improved patient diagnosis and care.
-Results integrated into the medical record.
Potential savings:
-Currently, millions of dollars spent on testing.
-10-25% savings from contract consolidation.
Requests for hires during initial pilot:
- Genetic Counselor $100,000
- Molecular Pathologist $220,000
- Administrative Support $50,000

CONCLUSION
• Molecular diagnostic testing is evolving rapidly with many options for patient testing.
• Robust patient counseling is essential for comprehensive medical care.
• Coordinated effort across the NIH campus can improve capabilities, patient care, and research potential.

References:

Presented at the 2019 ELAM® Leaders Forum
ABSTRACT: 2019 ELAM Institutional Action Project

Discussion/Conclusion: Molecular diagnostic tools improve our capability to diagnose complex and rare diseases and to make better treatment decisions. A proposal, with business case, was submitted to Clinical Center leadership for the following actions:

(1) Hire an expert in Clinical Molecular Genetics or Molecular Pathology to oversee molecular diagnostic testing within the Clinical Center.
(2) Create an advisory group of experts on campus, derived from those already working to address these issues, to advise the Clinical Center regarding testing practices.
(3) Develop a database of services, including costs of service and feedback on quality, as a resource for intramural investigators searching for testing options.