Assessing Success of a Quality Improvement Process for Tracking, Reviewing, and Correcting Constitutional Cytogenetic Test Orders in a Reference Laboratory

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INTRODUCTION

Incorrect genetic test orders delay diagnoses, provide false reassurance, and decrease the quality of patient care.

Utilization management (UM) by genetic counselors (GCs) decreases costs and improves patient care.

Rubenstein et al (2018) compared the efficacy of various clinical laboratory test UM practices; however, comparisons of UM processes specific to genetic testing have not been performed.

UM for postnatal constitutional cytogenetic test orders differs from many UM processes described due to high volume and limited clinical information provided with orders.

OBJECTIVE

To assess a quality improvement (QI) project for UM of postnatal constitutional cytogenetic test orders.

Goal of UM: To prevent redundant testing and ensure that the test(s) ordered are most appropriate given the suspected diagnosis.

Previous UM process: Support staff reviewed orders based on provided criteria and escalated cases not meeting conditions to GCs. See table 1 for examples of orders that would be flagged for GC review. Cases should be escalated to GCs by processors in the pre-analytical phase. Too often, cases were belatedly sent to GCs during the interpretation and report writing phases by medical directors and technologists.

Improved UM process: GC support specialists (GCSS) review orders, based on provided criteria, to determine whether GC involvement is necessary. A software program created for this purpose organizes and displays incoming orders.

GCs assess escalated cases by: provided clinical indication, previous and concurrent order history, and/or patient age and sex. GCs investigate then change, cancel or approve testing. GCs often update indications and add case-specific comments to reports.

If fewer orders miss escalation to GCs during the pre-analytical phase, the QI project is a success.

TABLE 1

Examples of cytogenetic testing orders which should be escalated to GCs
Indication is inappropriate for cytogenetic testing (e.g. hypothyroidism or hypertension)
Infertility evaluation with no karyotype ordered
Simultaneous FISH metaphase and cytogenomic SNP microarray orders
Orders with single gene testing indications (e.g. cystic fibrosis)
Orders for karyotype with microdeletion indications
All orders involving family history or previous testing indications

METHODS

August 2018 data from postnatal constitutional cytogenetic test order review, prior to the QI project, were pulled into a database and compared to August 2019 data, after QI implementation. Orders and indications for FISH (aneuploidy panel and single locus), karyotype, and cytogenomic SNP microarray were reviewed. A determination was made as to whether orders *should* have been escalated to a GC, and whether orders *had* been escalated.

Cases were stratified by indication: common (trisomy, malformation, autism, intellectual disability), infertility and multiple miscarriages, family history, other (egg/sperm donor or nonsensical), and follow-up testing.

The proportion of cases escalated to GCs and the proportion of cases that should have been escalated but were not, in each indication category, were calculated.

Constitutional test orders with oncology indications were not included due to separate protocols and processes.

Test volumes are not reported; however, statistical analysis was performed to evaluate the inferences and conclusions. P-values by a two-sample, two-sided test for equality of proportions were calculated.

RESULTS

Significant differences were identified (p< 0.001):

Prior to the QI process, 6% of postnatal constitutional cytogenetic test orders were escalated for GC review. An additional 9% should have been escalated but were not.

After the QI project, 18% of orders were escalated to a GC. An additional 2% should have been escalated but were not. There is a significant difference (p< 0.001) in both the % escalated and the % not escalated between 2018 and 2019.

Escalation of cases with common indications increased from 3% to 14%, with fewer (10% compared to 2%) cases not escalated.

Similar increases and decreases were seen for all indication categories except follow-up testing. See table 2.

TABLE 2

Indication Category	Orders in category / Total orders		Escalated to GC within category			Missed cases within category		
pre QI / post QI	August 2018	August 2019	2018 pre-QI	2019 post-QI	p-value	2018 pre-QI	2019 post-QI	p-value
Common	57%	65%	3%	14%	< 0.001	10%	2%	< 0.001
Infertility	32%	24%	0%	7%	< 0.001	4%	1%	< 0.025
Family History	3%	5%	79%	100%	< 0.002	21%	0%	< 0.002
Other	4%	2%	32%	38%	< 0.721	19%	0	< 0.062
Follow-up	2%	2%	25%	41%	< 0.295	5%	12%	< 0.425
Total			**6%**	**18%**	**< 0.001**	**9%**	**2%**	**< 0.001**

CONCLUSIONS

The process improvement has increased efficacy of postnatal constitutional cytogenetics utilization management, demonstrating an increase in the proportion of cases appropriately flagged for pre-analytical GC review, and a decrease in the proportion of cases that should have been escalated but were not.

References: Rubinstein M, et al. Effectiveness of Practices to Support Appropriate Laboratory Test Utilization: A Laboratory Medicine Best Practices Systematic Review and Meta-Analysis. *Am J Clin Pathol*. 2018 Feb 7;149(3):197-221.PMID: 29471324..

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