High throughput counseling: a model for genetic testing results disclosure and patient management

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Objective

Population-based carrier screening for single gene disorders has been a part of clinical practice for decades. The goal of preconception and prenatal carrier screening is to provide couples with information to optimize reproductive outcomes based on their personal values and preferences.¹ Historically, carrier screening was offered to individuals of particular ethnic backgrounds known to be at increased risk of certain disorders, known as ethnic-based screening. As ethnic distinctions become less defined, ethnicity-based screening has become less effective. With rapid advances in technology and decreases in cost of genetic screening in the past several years, it is now possible to screen for a large number of conditions simultaneously.² This testing strategy, known as expanded carrier screening (ECS), provides screening for the same expanded panel of disorders to all patients, irrespective of ethnicity. In 2017, the American Congress of Obstetrics and Gynecologists (ACOG) updated its recommendations to state that expanded carrier screening is an acceptable strategy for preconception and prenatal carrier screening.³ Utilization of ECS has increased the scope of information available to patients and providers alike. Along with this provision of information, guidelines highlight the importance of timely results delivery in the context of genetic counseling.⁴ Here we report on the results of a retrospective study of a large-scale automated results delivery system that includes tele-genetic counseling for patients undergoing expanded carrier screening.

Description of service

Guidelines from ACOG were utilized to develop patient notification, reminder, and tracking protocols. American College of Medical Genetics and Genomics (ACMG) guidelines and internal experts were utilized to create post-test education and counseling elements to develop a protocol for the delivery of carrier screening results. The overview of the Counsyl Complete⁵ results delivery system is illustrated in Figure 1. As shown, this workflow differs depending on result type.

Results

Over an eight-year period, 278,318 carrier screening results were issued through the automated results delivery system. The majority of the patient cohort was female (73.8%). Median age was 34 years (interquartile range (IQR): 30-37). Figure 2 shows the frequency of different result types in the patient cohort. A total of 43,343 consultations were performed during this study period for 41,050 patients (10%); 26% of these consultations were for screen-negative results and one-third (32%) of all consultations were on-demand. Median consultation time was ten minutes (IQR: 5-15 minutes), and median patient satisfaction rating for consultations was 4.95/5.

Conclusion

Combining web education, tele-genetic counseling, and automated notification protocols, we implemented a service that efficiently managed results disclosure for over 278,000 patients. This study demonstrates the feasibility of large-scale results delivery that includes both on-demand and scheduled tele-genetic counseling and yields high patient satisfaction. Automated results delivery platforms may help overcome barriers such as limited access to in-person genetic counseling in certain geographic areas, employment conflicts, long wait times for scheduling an in-person genetic counseling appointment, and appointment times outside of normal business hours. In summary, we show an efficient and scalable means of implementing medical guidelines on post genetic testing patient management in order to maintain high quality patient care. Scalable platforms such as the one described here will become increasingly important as genetic testing uptake grows among the general population.


Figure 2: Summary of results delivered through automated system

Figure 1: Automated Results Delivery Workflow

Figure 2: This figure shows the distribution of results delivered through the automated results delivery system by result type. Further, each result type is partitioned by the fraction of patients within the specific result type that elected a genetic consultation. Raw numbers and the total number of consultations per result type (as some patients had more than one consult) are shown in the table above.