Next-generation counseling: A model for non-invasive prenatal screening results disclosure and patient management

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Counsyl

Objectives

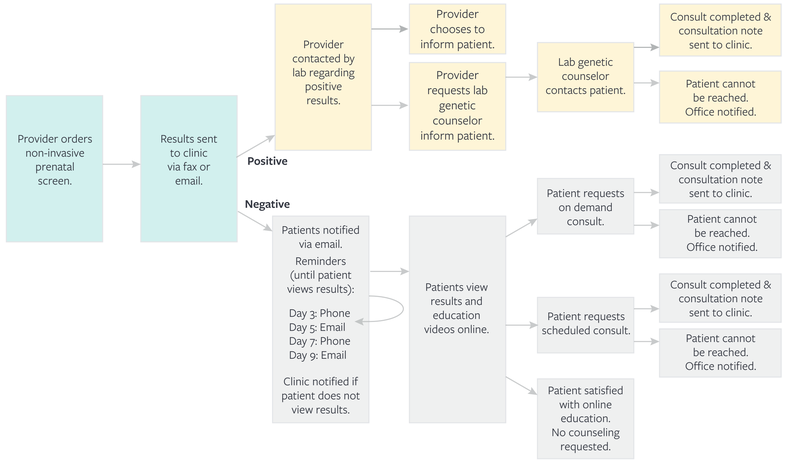
Non-invasive prenatal screening (NIPS) utilization has grown dramatically since its introduction, and is now increasingly being offered to the general population by non-genetics specialists.[1] Major guidelines recommend that patients with both positive and negative results should be counseled regarding limitations of testing, e.g., that the testing has nonzero false-positive and false-negative rates, and that an invasive test is required to confirm a positive result.[2,3,4,5,6] There is, however, a disconnect between these recommendations and the patient's clinical experience. Genetic counseling utilization is limited by, among other factors, inadequate supply of genetic counselors.[7] As a genetic testing laboratory that provides a results-delivery system (Counsyl Complete) that includes telecounseling, we report on how this service iis being utilized for patients undergoing NIPS.

Description of Service

Guidelines from the American Congress of Obstetricians and Gynecologists (ACOG) were utilized to develop patient notification, reminder, and tracking protocols while American College of Medical Genetics and Genomics (ACMG) guidelines and internal experts were utilized for the creation of post-test education and counseling elements in order to develop a protocol for the delivery of NIPS.[2,8]

The overview of the Counsyl Complete results delivery system is illustrated in **Figure 1.**

Figure 1: Automated Results Delivery Workflow



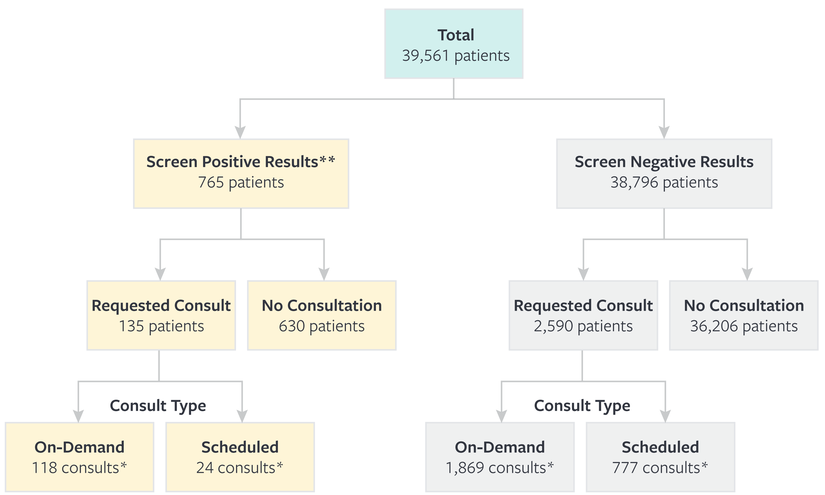
Upon results availability, providers are notified via fax, email, or electronic medical record (EMR). If negative, patients are contacted via an automated email and access results through a secure, HIPAA-compliant patient portal. Patients may watch tailored informational videos, request "on-demand" genetic counseling with a board-certified genetic counselor, schedule a later genetic counseling consult, or decline all of the above. Regardless of result type, if genetic counseling is elected, a consultation report is sent to the ordering provider via fax, email, or EMR.

Results

Over a 32-month period, 39,561 NIPS results (**Figure 2**) were issued through the system. Of these, 2,725 (7%) patients elected genetic counseling, 2,590 (95.1%) of which received negative results. 70.8% (n=1,834) of patients with negative results requested an on-demand consult while 83.7% (n=11) of patients with positive results requested an on-demand consult. Of the patients seeking a genetic counseling appointment, the average age was 34 years with a range of 16 to 47 years.

Average consultation time was 15 minutes (range: 4-54 minutes) for positive results and seven minutes (range: 1-40 minutes) for negative results. On average patients spoke with a board certified genetic counselor within 13 minutes when an on-demand consult was requested. The average patient satisfaction rating was 4.9/5.0.

Figure 2: Summary of Results



*Some patients had multiple consults

**Screen positive results include both aneuploidy suspected and aneuploidy detected

Conclusion

Combining web education, genetic telecounseling, and automated notification protocols, we implemented a service that efficiently manages NIPS results disclosure. The study demonstrates a desire for on-demand genetic counseling and showed high satisfaction with the service received, regardless of the type of result the patient received. This system 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.

We describe an efficient and scalable means of manifesting medical guidelines on post-NIPS patient management to a large number of patients, which is imperative to quality clinical care as uptake grows among the general population.

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