Objective: Over a 39-month period, 67,122 NIPS results (1,198 screen-positive and 65,924 screen-negative tests) were issued through the platform to 66,475 unique and eligible patients (Figure 1). Of this population, 4,655 (7.0% overall; 9.6% screen-positive) requested genetic counseling. NIPS results were delivered to the ordering provider, who may request that a laboratory-based genetic counselor contact the patient. All other results can be viewed directly by the patient in an online portal and are accompanied by written post-test education and counseling protocol for the delivery of NIPS and carrier screening results. Providers are notified upon availability of patient results.

For screen-negative results returning a single copy number ratio, patients may request an automated email directing them to schedule a consultation with a laboratory-based genetic counselor. Screen-positive NIPS results are delivered to the ordering provider, who may request that a laboratory-based genetic counselor contact the patient. All other results can be viewed directly by the patient in an online portal and are accompanied by written post-test education and counseling protocol for the delivery of NIPS and carrier screening results. Providers are notified upon availability of patient results.

Figure 3a: Carrier Screening Results Delivery: Upon results availability, providers are notified via fax, email, or EMR. If the result is screen-negative a consultation report is sent to the ordering provider via fax, email, or EMR. If positive, the provider will receive a phone notification and a consultation report via fax/email/EMR. If uncertain, a consultation report is sent to the ordering provider via fax, email, or EMR. The result is autoreleased and email notification is sent to the patient. If results are screen-positive, patients will only receive these results in the context of genetic counseling, unless otherwise instructed by the ordering provider. Should the patient request a consultation, a consultation report is sent to the ordering provider via fax, email, or EMR.

Figure 3b: NIPS Result Delivery: Upon results availability, providers are notified via fax, email, or EMR. If the result is screen-negative, a consultation report is sent to the ordering provider via fax, email, or EMR. If positive, the provider will receive a phone notification and a consultation report via fax/email/EMR. If uncertain, a consultation report is sent to the ordering provider via fax, email, or EMR. The result is autoreleased and email notification is sent to the patient. If results are screen-positive, patients will only receive these results in the context of genetic counseling, unless otherwise instructed by the ordering provider. Should the patient request a consultation, a consultation report is sent to the ordering provider via fax, email, or EMR.

Figure 3c: Ambulatory Results Delivery Overview: NIPS and carrier screening have dramatically widened the scope of information available to patients and providers alike, underscoring the importance of timely results delivery. We describe an automated system that utilizes a single workflow to deliver results and consistently deliver results to patients in a timely fashion and in the context of genetic counseling, adding that a policy of “no news is good news” does not represent high quality care. However, genetic counseling utilization is limited by, among other factors, an inadequate supply of genetic counselors.3

As the availability, uptake, and complexity of genetic testing expands in the general obstetrics setting, primary care providers increasingly must offer genetic services. Non-invasive prenatal screening (NIPS) and carrier screening have dramatically widened the scope of information available to patients and providers alike, underscoring the importance of timely results delivery. We describe an automated system that utilizes a single workflow to deliver results and consistently deliver results to patients in a timely fashion and in the context of genetic counseling, adding that a policy of “no news is good news” does not represent high quality care. However, genetic counseling utilization is limited by, among other factors, an inadequate supply of genetic counselors.3

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 genetic counseling. As illustrated in Figure 1, the majority of results delivered were screen-negative. All other results are classified as screen-positive.

Figure 3: 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 genetic counseling. As illustrated in Figure 1, the majority of results delivered were screen-negative. All other results are classified as screen-positive.

Conclusion: Over a 39-month period, 67,122 NIPS results were issued, of which 1,198 were screen-positive. Of the NIPS results delivered, 4,655 (7.0% overall; 9.6% screen-positive) requested genetic counseling. Of the screen-negative results delivered, 6,654 (9.9% overall; 10.3% screen-negative) requested genetic counseling. 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 genetic counseling. As illustrated in Figure 1, the majority of results delivered were screen-negative. All other results are classified as screen-positive.

REFERENCES

Figure 2: 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 genetic counseling. As illustrated in Figure 1, the majority of results delivered were screen-negative. All other results are classified as screen-positive.

NIPS Results: As illustrated in Figure 1, the majority of results delivered were screen-negative. All other results are classified as screen-positive.

Carrier Screening Results: As illustrated in Figure 1, the majority of results delivered were screen-negative. All other results are classified as screen-positive.

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REFERENCES