

## I. Introduction

Genetic Results Reporting (RR) services were developed by our group to increase patient access to genetic counseling expertise and provide support to referring providers (RPs). Referring providers authorize our genetic counselors to contact patients with abnormal results via telephone. Patients are called by a genetic counselor to discuss positive carrier screening or positive or failed cell-free DNA test (cfDNA) results. An interpretation service is used when needed to manage any language barriers. Genetic Results Reporting is a focused discussion of the results, including an overview of the disorder for which the patient is a carrier or the fetus is at increased risk, and a discussion of follow up testing options. The purpose of this study is to determine the effectiveness of a results delivery model provided by genetic counselors.

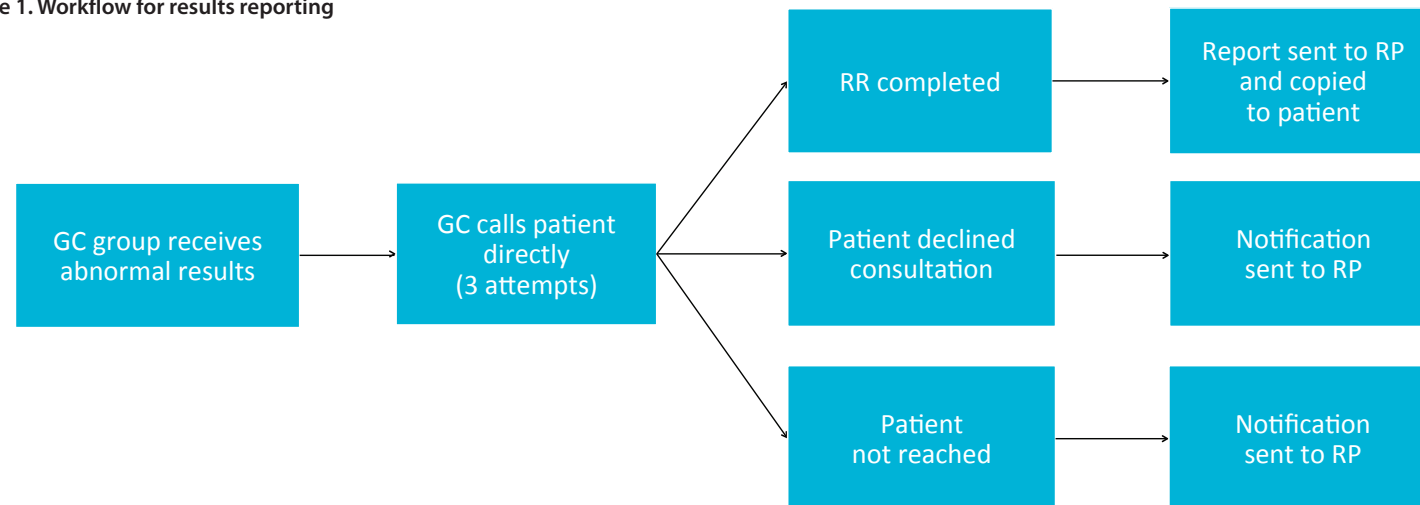
## II. Methods

Referring providers are instructed to inform patients that a genetic counselor from our group will contact them if their result is abnormal. To encourage patients to answer calls from unknown phone numbers, the provider offices are given postcards with a description of the program to provide to their patients.

Our group is alerted to abnormal results when result is released from the laboratory. Genetic counselors call the patient on that day and on the next two consecutive work days if the patient is not reached on the first attempt. To ensure that patients are informed of abnormal results in a timely way, referring providers are informed by phone and fax when we were not able to reach their patient by the third day. When patient contact is successful, a report is sent to the referring provider documenting the notification of abnormal results.

Data was reviewed for 31 months, from May, 2018 to November, 2020 to determine if Genetic Results Reporting was completed for patients who had positive results, if the patient declined a Genetic Results Reporting discussion with the genetic counselor, or if we were unable to contact the patient.

Figure 1. Workflow for results reporting

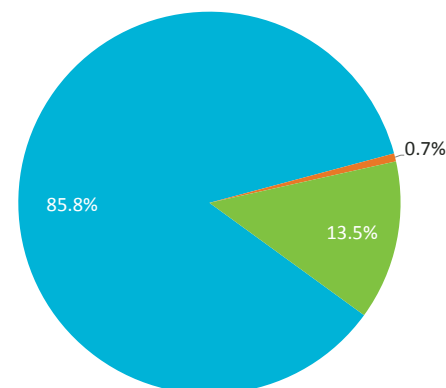


## III. Results

During the study period, a total of 6,483 patients qualified for the program. We were successful in contacting 5609 (86.5%) of patients. Upon reaching patients, a small number (44) of patients declined a results discussion; most often because they already were aware of their cfDNA results or their carrier status. The final success rate of completing Genetic Results Reporting with patients with positive carrier screening or cfDNA results was 85.8%.

Figure 2. Success rate of results reporting

■ Complete  
■ Declined  
■ Did not reach



## IV. Conclusions

Genetic Results Reporting was shown to be a highly effective approach to inform and provide patients with information about their carrier screening or cfDNA results. Given the increasing complexity of genetic testing results, referring providers may have limited time to provide appropriate counseling. Support from genetic counselors can provide referring providers more confidence in offering complex genetic tests to their patients. Our study demonstrated that our model of Genetic Results Reporting is an effective method to increase patient access to a genetic counselor's expertise.