

# Patient-friendly Reports: Incorporating Genetic Counselors into the Report Development Process

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## Introduction

Demand for genetic testing is growing, with an expected \$25 billion to be spent on genetic testing in the United States by 2021, up from \$5 billion in 2014<sup>1</sup>. This growth is causing increased expectations on health care providers, who may themselves have only a basic knowledge of genetics. Additionally, explanation of genetic concepts can take up much of the limited time a provider has to spend with a patient. In some instances, misunderstanding of report information has led to adverse events, such as inaccurate diagnoses or unnecessary surgeries<sup>2</sup>, highlighting the need for increased clarity in genetic testing reports for both health care providers and patients. Potentially, such mistakes could be mitigated if patients were able to understand their own reports and engage in more informed discussions with their providers. However, it is challenging to create a report that is understandable for someone with a low health literacy level, yet maintain informational accuracy.

Genetic counselors are trained to describe complex ideas in easy-to-understand terms. Therefore, genetic counselors can be a helpful addition to the report development process by writing reports in a way patients can comprehend. There has been some research on how to create patient-friendly report layouts<sup>3</sup>, but nothing to date regarding how to use genetic counselors in creating a patient-centered report. Here, we describe our approach for creating patient-friendly reports in a commercial laboratory setting and patients' perceived clarity of results.

## Methods

At the beginning of the report development process, literature curation is performed by a team of genetic counselors under the direction of a medical geneticist. Members of the curation team search PubMed for relevant information using a specific set of parameters agreed upon by the team at the beginning of the project. Search parameters may include the gene name and key terms such as "cancer risk", "odds ratio", or "meta-analysis". For new reports, the curation team writes a first draft of new content based on findings from the literature review. In order to keep all content current, the curation team searches for and reviews new literature every six months. The team then recommends updates to references and report information when applicable.

When curation is complete, the first draft of new or updated content is reviewed by the content team, which consists of genetic counselors, a scientific writer, and a medical geneticist. The content team revises the draft, and when the team reaches a consensus, the content is passed on to additional reviewers. Additional reviewers may include external leaders in the medical field who have experience related to the content, members of the internal legal team, and members of the internal product and design team. These reviewers suggest edits to the content based on their individual perspectives. For example, design professionals contribute their expertise to generate patient-friendly layouts and visual aids, and experts in technology generate interactive, secure, web-based reports in addition to static reports standard in the industry. All edits suggested by the additional reviewers are assessed and accepted or rejected by the content team. The lab director reviews and formally approves the report content and releases it to the software engineering team to be implemented. Once reports are live, user feedback surveys are given to patients to allow them to provide opinions on what they like and dislike about the reports and suggest improvements. This user feedback is reviewed and incorporated into future versions of the reports.

For this study, we evaluated results of the user feedback survey. The responses of 4446 patients who underwent genetic testing through Color between 11/1/2015 and 2/28/2018 were included in the analysis. As part of the survey, users were asked to rate the statement "How clear were your results?", where a rating of 1 indicated they felt their results "were very confusing" and a rating of 5 indicated "I understood everything perfectly".

## Results

Figure 1. Report generation workflow

Stratified by personnel role. Additional reviewers include external specialists, legal, product, design, and lab director. + indicates the report has passed that stage and can proceed, - indicates the report has failed that stage and must return to a previous stage.

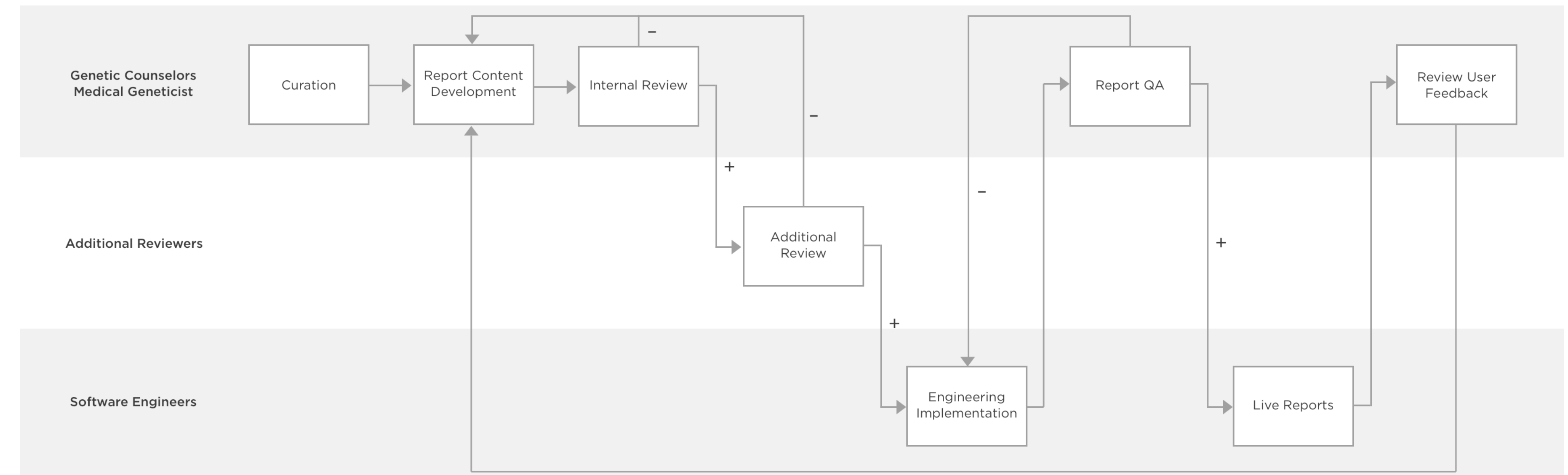


Figure 2. Positive sample report

Results are returned via interactive online reports and static PDFs for printing and faxing. Screenshots of the online version of a positive report are shown below.

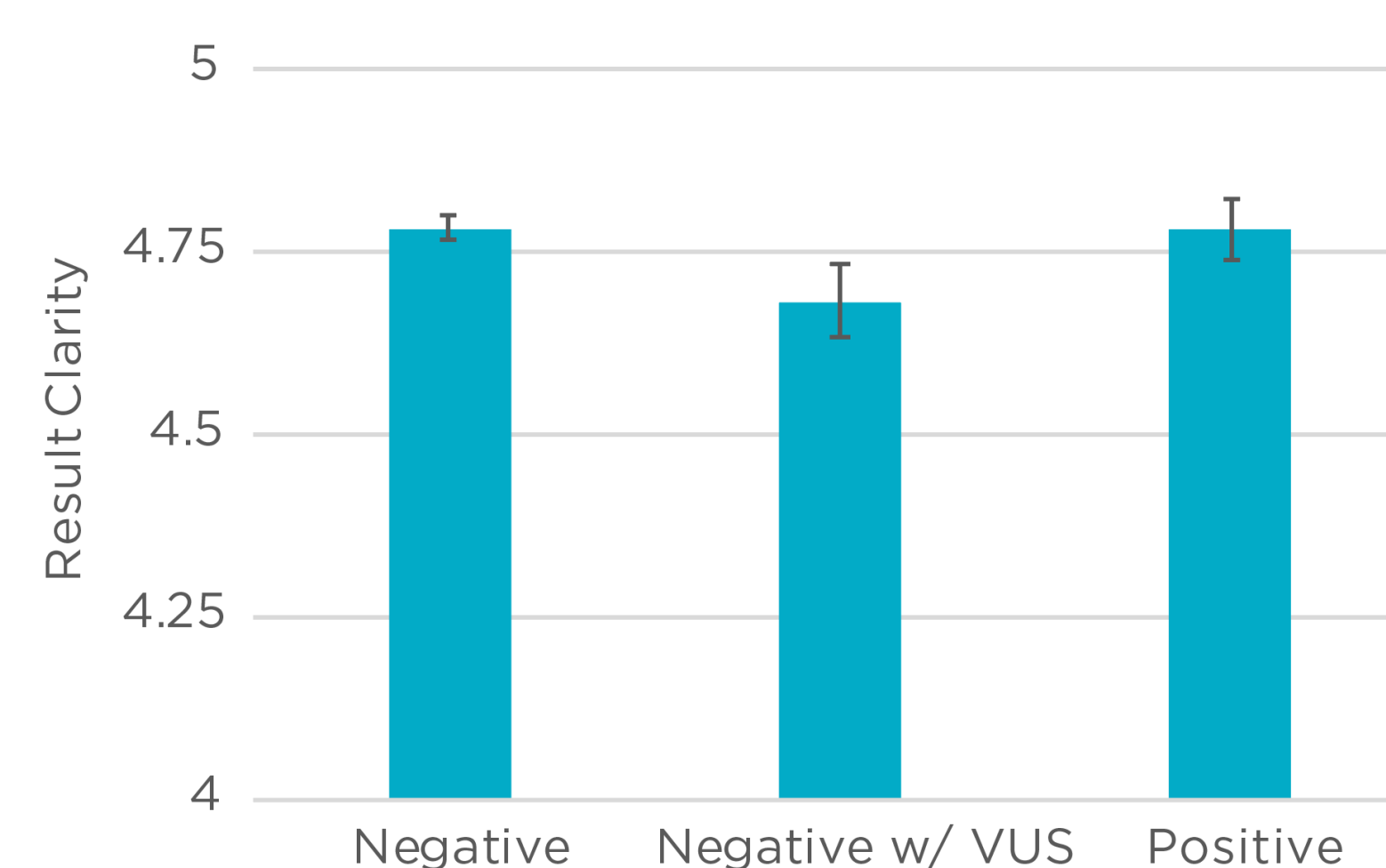
**1** Patient's personal history is highlighted in the opening summary (if applicable), and patient is encouraged to relay their history to their healthcare provider.

**2** Cancer risks are clearly illustrated in an easy-to-read and interactive format, which teaches patients and reminds providers about the specific risks associated with pathogenic variants in that gene.

**3** Recent screening guidelines from the National Comprehensive Cancer Network (NCCN) are provided as **Next Steps** in the report for patient and provider reference.

Figure 3. Survey results

Answers to "How clear were your results?". Results were reported positive if one or more pathogenic or likely pathogenic variant was detected, negative if no variant and/or only benign or likely benign variants were detected, and negative with VUS if a variant of uncertain significance was detected. Mean response, +/- 95% CI.



## References

- <sup>1</sup> United Health Working Group. 2012.
- <sup>2</sup> Bonadies DC, Brierley KL, Barnett RE, et al. *Cancer J*. 2014.
- <sup>3</sup> Haga SB, Mills R, Pollak KI, et al. *Genome Med*. 2014
- <sup>4</sup> Scherr CL, Lindor NM, Malo TL, Couch FJ, Vadaparampil ST. *Clin Genet*.

**4** The **Details** section includes information for the patient about the affected gene and its function.

**5** Also included is relevant information for providers about the specific variant.

**6** In the **Questions** section, genetic counselors created a list of common questions to address within the report based on real questions they had often been asked during counseling sessions.

Traditional genetic counseling information such as the probability that certain relatives have also inherited the variant and who should consider testing is included in the **Family** section.

## Discussion

- In our experience, genetic counselors can be a helpful addition to the report development process in order to make reports patient-friendly without losing informational accuracy.
- Genetic counselors can help provide information that is typically covered in a counseling session for patients while also ensuring information relevant to providers is highlighted in an appropriate way.
- When asked about the clarity of their results, users responded above 4.5 out of 5 regardless of result type. The slightly lower clarity rating amongst those with a VUS is expected, as the concept of a VUS has traditionally been considered difficult for genetic counselors to communicate to patients<sup>4</sup>.
- Patient friendly reports aim to empower patients to be their own advocate and make informed medical decisions.