

Designing for inclusivity in population genomics

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Introduction

Advancements in genetic testing and technology have paved the way for personalized medicine, with promises of providing more equitable and tailored care. Services and tools that make genetics more universally accessible and context specific are increasingly important as genomics programs and genetic testing are available to larger and more diverse populations.

The issue of gender equity is particularly relevant in the realm of population genomics. It is essential that 1) people feel welcomed and respected as they onboard into the process, 2) the lab gets the information that it needs to ensure that samples are not mis-attributed, 3) the lab has the information it needs to make sure that health risks are communicated effectively, and 4) all patients trust that the information they are receiving is accurate for their body and their health.

Historically, Color's Hereditary Cancer Test results reported binary risk estimates and screening guidelines derived from analyses of large populations of individuals based on sex determined at birth without consideration of gender identity. This information, while generalizable for some, may be clinically inaccurate, misleading, and disrespectful for those who identify differently than their sex determined at birth.

To improve our participant experience, we implemented a health history intake workflow which enables non-binary and transgender individuals to identify their preferred gender identity in addition to annotating their sex determined at birth. By collecting these unique pieces of information, we can provide gender affirming care through personalized genetic test results and resources. This implementation has also equipped our clinical team, including genetic counselors, with valuable background information that accounts for an individual's identity as well as their specific risks and medical recommendations. Additionally, to expand on the investment to provide client-centered care, Color's genetic counseling team has begun conducting training on providing gender affirming genetic counseling for these patients.¹ Here, we describe the participant experience, the quality control mechanisms, and the process of providing clinical reports to non-binary and transgender individuals.

Methods

To increase inclusion, we implemented changes within our online activation and health history intake to allow individuals the option to self-identify their preferred gender identity in addition to their sex determined at birth. Clients who opt to provide more information about their gender can select one of the following: female, male, non-binary, prefer not to say, or prefer to self-describe. Throughout this process, clients are also provided information on how these answers inform their experience and results. For example, clients are notified that Color tests obtain information generated according to an individual's chromosomes (typically in line with sex assigned at birth) and that the gendered content within a report may not match their identity.

Separately, a quality control (QC) metric, which estimates the number of X and Y chromosomes in a sample, was implemented to ensure reporting accuracy. When there is evidence of a discrepancy (i.e. predicted XX and male reported sex), our clinical team is prompted to investigate the cause. Though there are several potential reasons for a mismatch, this metric specifically brought to light several cases of discrepancy due to differences between a client's reported gender identity and chromosomal sex. This process further highlighted opportunities to better represent our non-binary and transgender clients within our health history intake and reporting workflow.

Upon report generation, individuals who note a gender identity that is different than their sex determined at birth or their chromosomal sex determined by QC metric are provided personalized reports that acknowledge this difference and an explanation for the report content not reflecting their noted gender identity.

Qualitative feedback from Color clients and genetic counselors and quantitative data were collected prior to and after implementation. All individuals consented to have their de-identified information used in anonymized studies. All demographic information was reported by the individual.

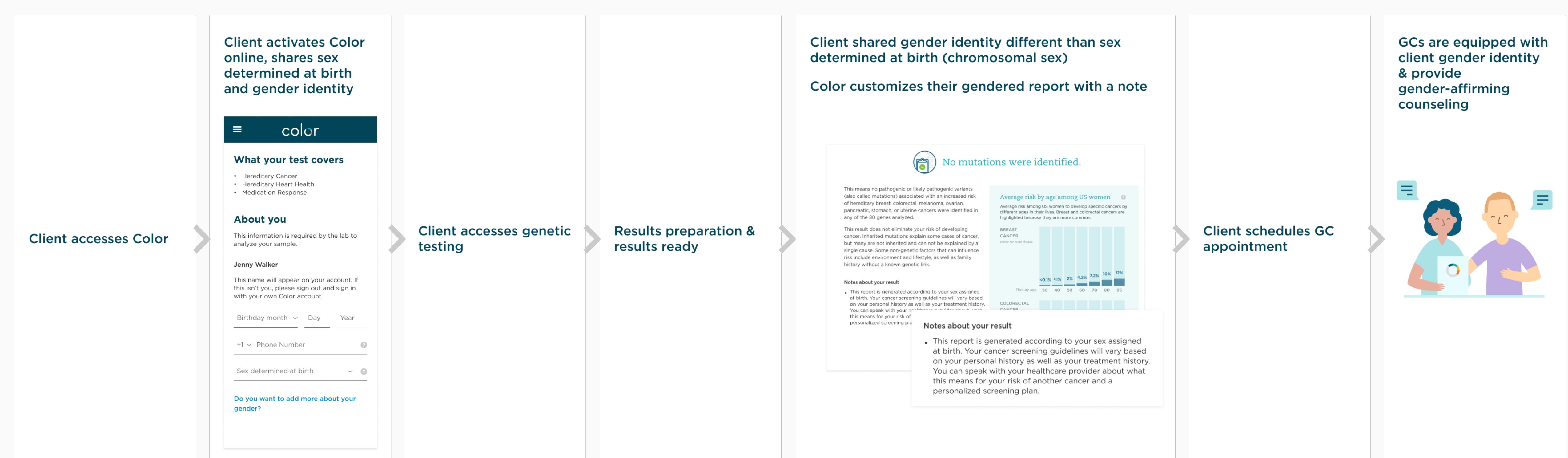
Conclusions

- Non-binary and transgender individuals seeking genetic testing information appreciate the ability to self-identify their preferred gender identity and sex determined at birth.
- Implementing changes within our health history intake workflow to collect this information supports gender affirming care through personalized genetic test reports and resources.
- Knowledge of a client's gender identity and sex determined at birth equips GCs to provide supportive counseling personalized to the individual's health needs.

Results

Figure 1. Process workflow

Customized workflow to promote a more personalized genetic testing experience for our non-binary and transgender clients.



Multiple entry points: Client completes health history intake.

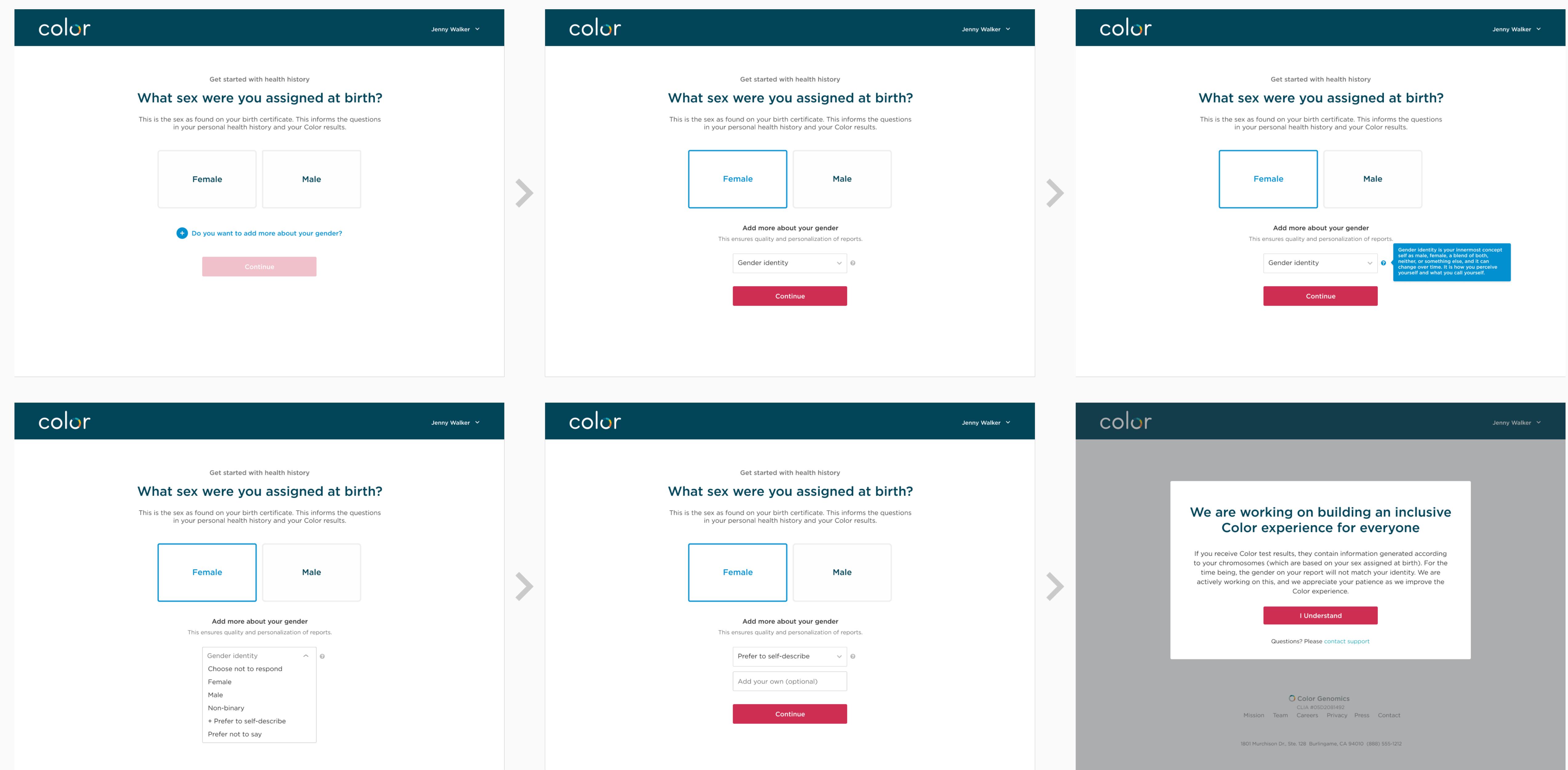


Figure 2. Qualitative data prior to workflow changes

Prior to workflow changes, Color clients expressed a desire to incorporate their gender identity into their genetic testing and reporting experience.

- "I am Male to Female transgender and wondering how to answer a lot of questions on the history section. I've selected Female for sex but some of these questions don't apply (never had a period, for example). I couldn't go back to see if the Male section would be more applicable, but that also wouldn't feel correct."
- "There we no place to put comments but I thought these confidential comments are important to analyzing my results. I am a MTF transgender woman and I take Estrogen. So, besides having a male anatomy, I also have a woman's breasts. My legal gender is Female. MTF transwoman...Might I suggest that you add the ability of someone to indicate they are transgender..."
- "I am a transgender male, not cisgendered."

Figure 3. Qualitative data after workflow changes

After implementation of workflow changes, clients and GCs noted the positive outcomes.

- After one of our female to male transgender clients received a report with screening guidelines and risk estimates relevant for female sex determined at birth, he cited this type of report as "most meaningful" to him. Our Color GC noted he was appreciative of the acknowledgement and work towards becoming more inclusive.
- With knowledge of the client's gender ID and reported sex determined at birth, our Color GC was able to counsel a female to male transgender client specific to his healthcare needs, with particular care in discussing risks and screening options as it related his history.

References

1. Zaykowski, K. et al. J. Genet. C of Genet. Counsel. (2019).