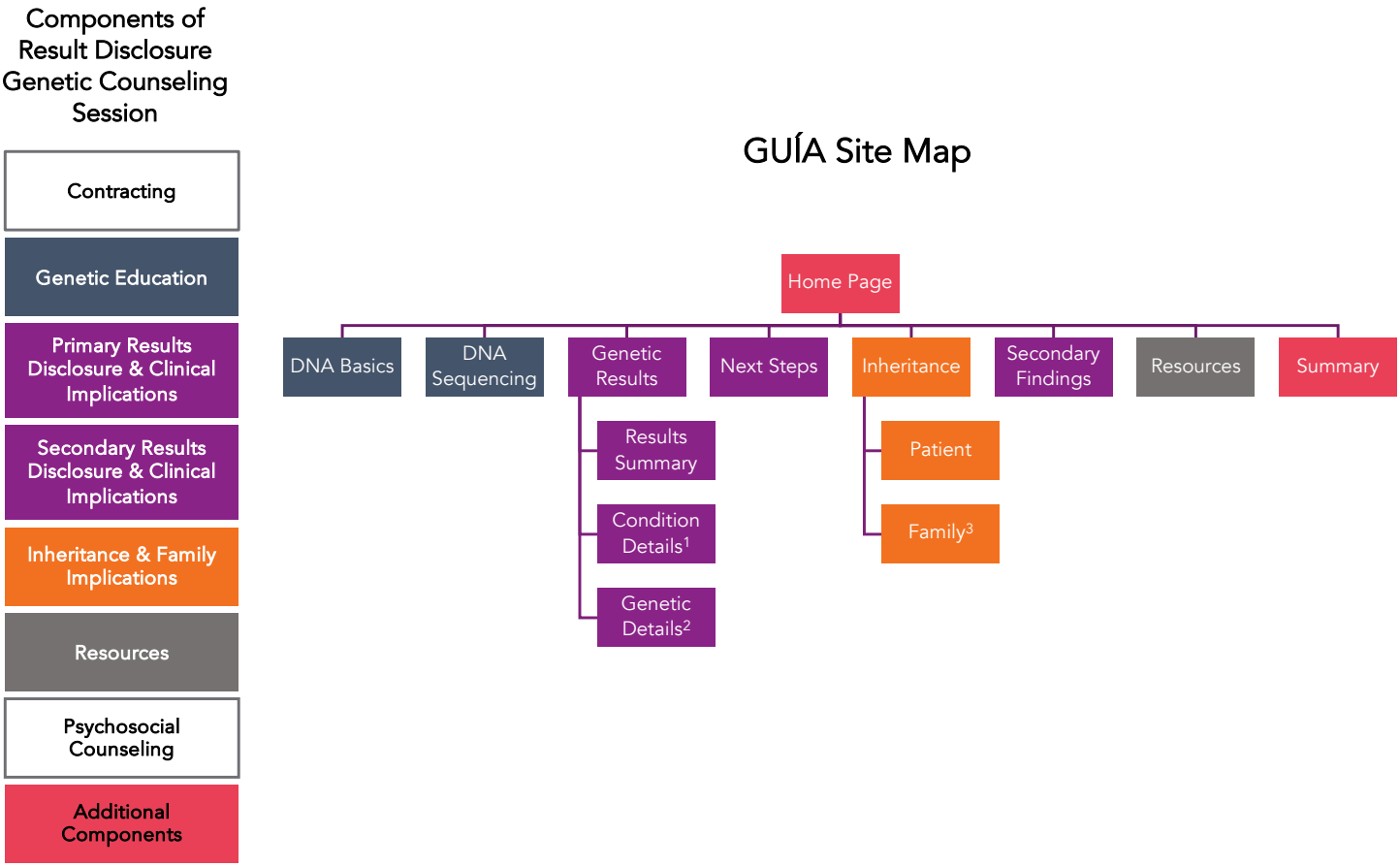
Supplementary Materials

GUÍA: a digital platform to facilitate result disclosure in genetic counseling

Supplemental Figure 1. GUÍA pages mapped to the components of a genetic counseling result disclosure session. Components of the genetic counseling session are displayed in the same color as the corresponding GUÍA page or subtab.

Supplemental Table 1. GUÍA content colored to map to the GUÍA site map found in Supplemental Figure 1.

Supplemental Table 2. Examples of input from the Genomics Community Board and implications for GUÍA design and development



Supplemental Figure 1. **GUÍA pages mapped to the components of a genetic counseling result disclosure session.** Components of the genetic counseling session are displayed in the same color as the corresponding GUÍA page or subtab.
[1]Displayed for positive results only
[2,3] Displayed for positive and uncertain results only

Supplemental Table 1. GUÍA content colored to map to the GUÍA site map found in Supplemental Figure 1.

Web page	Result category	Content
Genetic Education		
DNA Basics	All	• Explanation of what a gene is and what genes do in the body • Explanation of what DNA is and what the function of DNA is in the body • Description of a genetic variant
DNA Sequencing	All	• Explanation of the type of genetic test performed and why • Description of how DNA sequencing works • Possible results: positive, negative, uncertain, secondary findings
Primary Results and Clinical Implications		
Results	All	• Clinical interpretation of the results and explanation of what the results mean • Possibility for variant reclassification[1]
Condition Details	Positive	• Description of the associated condition and symptoms • Images of the body systems affected by the condition • Link to condition specific resource(s)
Genetic Details	Positive, Uncertain	• Table displaying variant information: gene, genomic position, zygosity • Description of gene function • Link to gene specific resource(s)
Next Steps	All	• Clinical management recommendations (additional testing, treatment, referrals) • Link to resources (clinical trials)
Inheritance and Family Implications		
Patient	All	• Description of the inheritance pattern of the condition[2] • Image showing inheritance pattern for the condition[2] • Reproductive risk for patient • Link to information about genetic inheritance
Family	Positive, Uncertain	• Reproductive risk for patient's parents[3] • Image displaying inheritance pattern personalized for the patient[2] • Recommendations for genetic testing of other family members
Secondary Findings, Clinical Implications, Family Implications		
Secondary Findings	Positive and negative secondary findings	• Description of secondary findings • Family's decision to opt-in or opt-out of receiving secondary findings • Clinical interpretation of results • Link to information about secondary findings
	Positive secondary findings	• Description of the associated condition and symptoms • Table displaying variant information: gene, genomic position, zygosity • Description of the gene function • Link to gene specific resource(s) • Description of the inheritance pattern of the condition • Reproductive risk for patient and patient's parents • Recommendations for testing of other family members • Clinical management recommendations (e.g., additional testing, screening, treatment, referrals to specialists) • Links to information about condition, gene, inheritance
Resources		
Resources	All	• Genetic counselor contact information • Links to additional resources
Summary		
Test Summary	All	• Clinical interpretation of primary results • Table displaying variant information: gene, genomic position, zygosity • Clinical management recommendations for primary positive or uncertain results • Clinical interpretation of secondary results • Genetic details for positive secondary results • Clinical management recommendations for positive secondary results • Genetic counselor contact information • Links to relevant resources

[1] Displayed for uncertain results; [2] Displayed for positive results; [3] Displayed for negative results; [4] Displayed for uncertain and negative results on the patient page

Supplemental Table 2. Examples of input from the Genomics Community Board and implications for GUÍA design and development

Genomics Community Board Input	Implication for GUÍA Design
Use images to help explain complex concepts • Biology textbook type images are images that people are familiar with and may be a helpful trigger	**Consulted medical illustrators on the design of the illustrations**
Results labeled as "negative" can be misunderstood to mean there are negative implications for a patient's health • Define what negative result means with the phrase 'no genetic cause found'	**All result categories are defined**
Secondary finding label does not have the same meaning for patients as it does for genetics professionals • Consider using another description, such as "other genetic results"	**Used the term 'secondary findings' for consistency across all study documents. The term is defined and there is a link provided to access additional information about this category of results**
Patients can have limited understanding of the results and may lack social support • Include resources and consider connecting with a social worker who understands genetics	***Resources* page can be personalized depending on the needs of the patient-participant**
Dark shading of the affected individual in an inheritance pattern illustration is not appropriate and potentially offensive • Consider using colors, patters, or other methods to identify the affected individual	**Color and labels were used to differentiate the affected and unaffected individuals in inheritance images**
Be thoughtful about the amount of text displayed • Simplify the *Home Page* by removing text include in early iterations of the design	***Home Page* contains only stand-alone titles**
Should not expect patients to remember the type of testing performed • Remind patient of the testing that was performed	***Learn About Sequencing* page includes a reminder of the type of test performed and why; *Secondary Findings* page includes a reminder of opt-in/out decision**
Organ system images may be confusing • Consider displaying organ system illustrations within a body, or circling the affected organs	Due to development limitations we could not implement this suggestion