GENEtest-101:

Developing an App for Healthcare Providers Working with Rare Disease Patients

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National Human Genome Research Institute

The Forefront of Genomics®

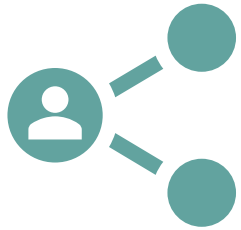
Project Overview

- **Goal:** Create a tool that could be utilized by healthcare providers as a point-of-care resource throughout the genetic testing process
- **Audience:** Pediatricians, family physicians, PAs, NPs, etc.
- **App link:** tinyurl.com/genetestapp
  - Please use/share!

Stage 1: PAS Workshop 1-pager and resource list

Stage 2: App Prototype Development

Stage 3: App Evaluation and Revisions

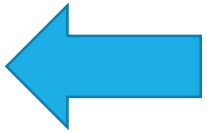


Stage 4: App Dissemination

Project Overview

PAS Workshop Materials

Toolkit 1-pager



Training Next Generation Pediatricians in Genomics: A Case-Study Approach

Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)

Initial Clinical Evaluation

ID "Red flags" that suggest a patient could have a rare genetic condition and may need a referral to genetics:

- Utilize Family GENES acronym (Australian Medical Association Genetic and Rare Disease Network)
- When to Suspect a Genetic Syndrome (*American Family Physician*)

As appropriate, refer patient to a genetics professional or proceed with genetic testing:

- Find a Genetics Clinic (ACMG)
- How to Find a Disease Specialist (GARD)

Test Selection

- Genetic Testing 101: Genetic Testing Topics (MedlinePlus Genetics)
- Determine appropriate genetic test(s): Comparing Genetic Tests (Jackson Lab), Genomic Diagnosis for Pediatric Disorders (*Frontiers in Pediatrics*)
- Select genetic testing laboratory: Genetic Testing Registry (NCBI)
- Consider potential follow-up studies

Pre-test Counseling

- Explain genetic testing options and facilitate patient decision-making: Genetic Testing FAQ (NIH NHGRI), Counseling About Genetic Testing and Communication of Genetic Test Results (ACOG)
- Obtain informed consent for testing: Informed Consent and Pretest Counseling Checklist (Jackson Lab), What is Informed Consent? (MedlinePlus Genetics)
- Ethical/Legal Considerations: Genetic Information Nondiscrimination Act (Jackson Lab), Genetic Testing of Children (AMA)

Diagnostic Result

- Interpret genetic test results: Utilize variant nomenclature (Human Genome Variation Society), ACMG-AMP sequence variant interpretation recommendations (*Genetics in Medicine*), and resources such as ClinVar (NCBI) and Franklin (Genoox) to assess variant pathogenicity (particularly for variants of unknown significance)
- Explore variant clinical relevance using resources such as ClinGen (NCBI), GeneReviews (NCBI), and OMIM (Johns Hopkins)
- Consider whether follow-up genetic testing may be helpful: parental testing (de novo vs. inherited), broader testing if results are negative (such as WES/WGS)
- Refer to genetics professionals for complex or uncertain results: Find a Genetics Clinic (ACMG)

Explain Results to Family

- Contracting: Ask family about their current understanding and for any initial questions. Describe the purpose of visit: (Genetic Counseling Toolkit)
- Disclose results: Test Results (Genetic Counseling Toolkit), What Do the Results of Genetic Tests Mean? (MedlinePlus Genetics)
- Address psychosocial concerns: Caring for a Patient with a Rare Disease (GARD)
- Integrate into clinical care: Utilize GeneReviews article if available to communicate screening, management plans, genetic risks to other family members, and reproductive options

Next Steps

Connect families with rare disease resources and/or condition-specific support groups:

- Info: GARD, NORD, Genetic Alliance, MedlinePlus Genetics
- Support: Family Voices, Parent to Parent USA, Facebook Support Groups
- Research: Undiagnosed Disease Network, clinical trials/research studies

Excel Resource List

Training Next Generation Pediatricians in Genomics: A Case-Study Approach (Inter-Society Coordinating Committee for Practitioner Education in Genomics)

Resource	Organization	URL	When to use it					What is it?	Tips and Tricks
			Clinical Eval	Test Selection	Pre-test	Diagnostic Result	Explaining Results		
MedlinePlus Genetics	NIH U.S. National Library of Medicine	https://medlineplus.gov/genetics/					INFO	Patient-friendly info about genetic concepts, conditions, and testing options. Printable PDFs of all content.	This site is a good first place to go if you are unfamiliar with a particular condition or concept. Search for specific conditions to find info in patient-friendly language.
OMIM	Johns Hopkins Medicine	https://www.omim.org/					INFO	Provides info about gene variants and associated phenotypes with helpful references and links to other sources.	Use orange "Clinical Synopsis" button to view summary of associated features; this can be used to help determine differential diagnosis. List on the right side of the page links to other helpful resources.
GeneReviews	NIH U.S. National Library of Medicine	https://www.ncbi.nlm.nih.gov/books/NBK1116/					INFO	Point-of-care resource that provides expert review articles about genetic conditions, covering diagnosis, management, and genetic counseling.	Differential diagnosis section provides list of conditions with overlapping signs and symptoms. Helpful determining conditions to consider. Genetic counseling section discusses recurrence risks.
PubMed	NIH U.S. National Library of Medicine	http://www.ncbi.nlm.nih.gov/pubmed					INFO	Database of biomedical primary literature reports and manuscripts. Helpful for finding primary literature related to specific biomedical topics; helpful if other sources such as GeneReviews don't have sufficient info.	Use filters on the left side of the webpage to limit search results to recently-published papers or certain article types such as review articles.

App Development

- Created using Google slides template and optimized for use on mobile devices
- Adapted content from toolkit to address the genetic testing process
- Included instructions for users to add to phone home screen
- Converted toolkit excel database into a section called “Resource List”
- Added links to download pdf and/or excel documents

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Explaining Results

The process of explaining test results can vary depending on the results themselves and the individual patient. Regardless, using patient-friendly language and addressing psychological issues that arise is an important component of results disclosure.

- Explaining positive results
- Explaining negative results
- Explaining uncertain results
- Using patient-friendly language
- Addressing psychosocial concerns

View resource list

Explaining Negative Results

While for some patients and families negative results may be a relief, for others it may mean that they are still without an explanation for their symptoms. In those cases, guidelines for explaining positive results can be helpful for framing the disclosure. Other key points to keep in mind include:

- A negative result does not guarantee that there is no genetic cause of the features seen.
- Follow up testing may be indicated, depending on the patient's features and what testing was previously performed.
- In the future, reanalysis or retesting may be able to provide a genetic explanation. Likelihood of this depends on the overall likelihood of a single genetic cause for the patient's symptoms.

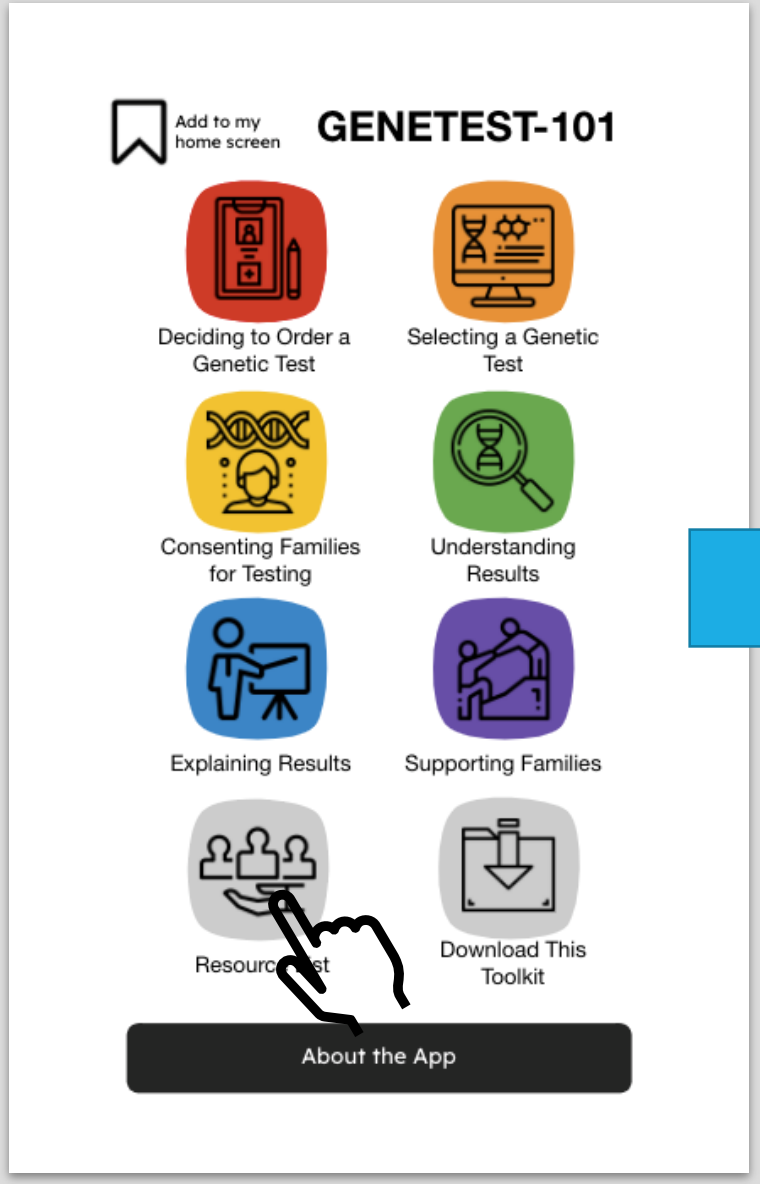
PREVIOUS: Explaining positive results NEXT: Explaining uncertain results

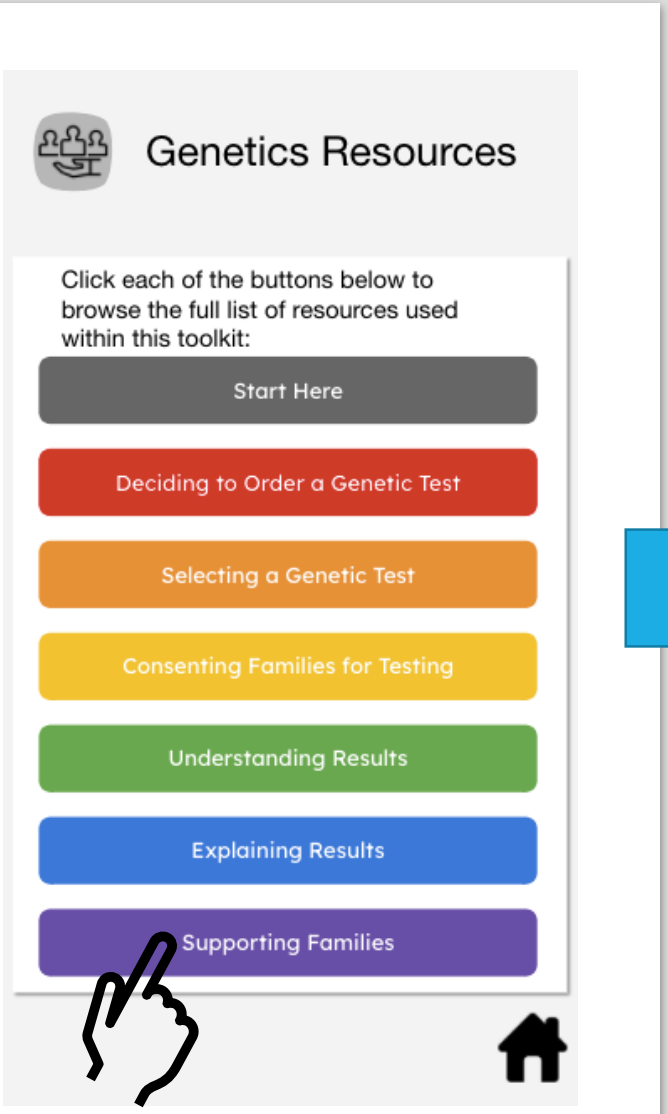
Further Evaluation of Variants of Uncertain Significance, cont.

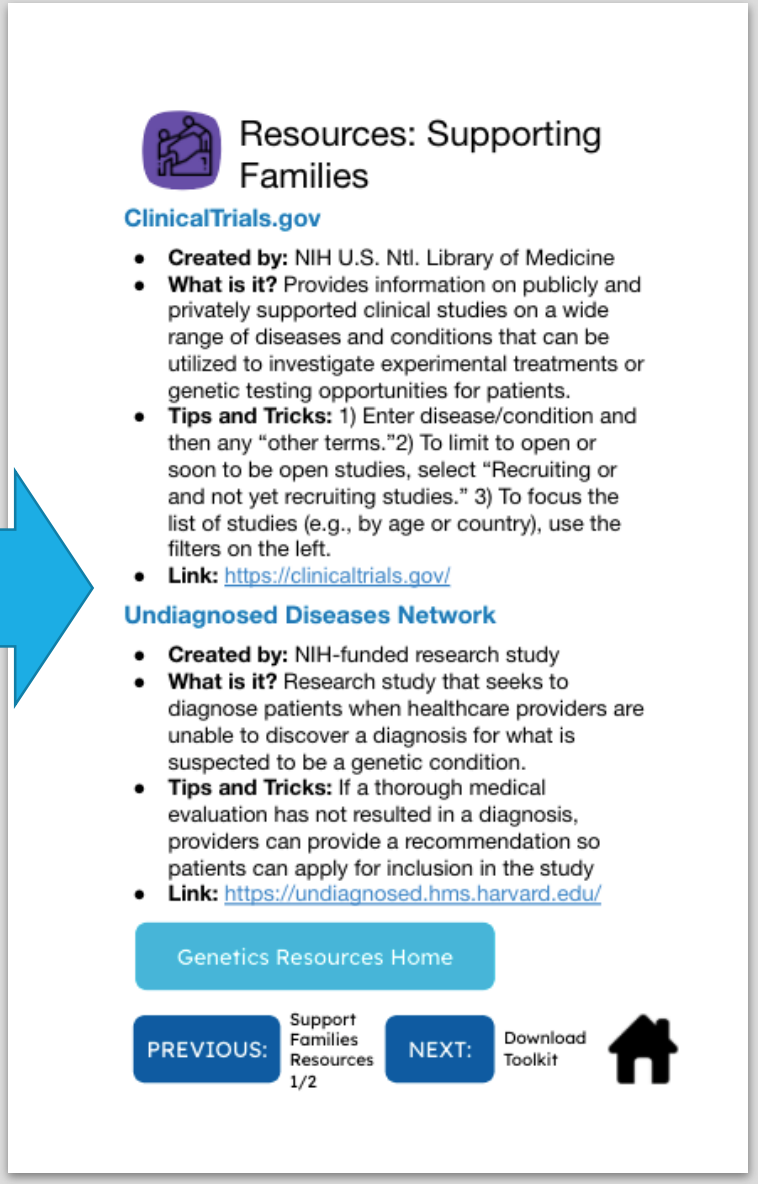
Follow-up Testing: If test results were negative and the patient's symptoms strongly suggest an underlying genetic cause, broader testing can be considered. This may involved a chromosomal microarray (CMA) to investigate deletions and duplications across the genome. It may involve whole exome or whole genome sequencing to investigate sequence changes in a broader, less targeted fashion.

If test results are uncertain, complex, or you are unsure if follow-up testing should be conducted, referral to a genetics professional may be indicated.

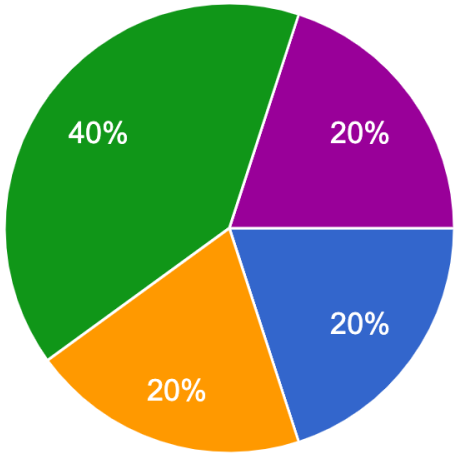
PREVIOUS: Evaluate VUSs 2/2 NEXT: Explaining results


Add to my home screen
GENETEST-101
Deciding to Order a Genetic Test
Selecting a Genetic Test
Consenting Families for Testing
Understanding Results
Explaining Results
Supporting Families
Resources
Download This Toolkit
About the App


Genetics Resources
Click each of the buttons below to browse the full list of resources used within this toolkit:
Start Here
Deciding to Order a Genetic Test
Selecting a Genetic Test
Consenting Families for Testing
Understanding Results
Explaining Results
Supporting Families


Resources: Supporting Families
ClinicalTrials.gov
Created by: NIH U.S. Ntl. Library of Medicine
What is it? Provides information on publicly and privately supported clinical studies on a wide range of diseases and conditions that can be utilized to investigate experimental treatments or genetic testing opportunities for patients.
Tips and Tricks: 1) Enter disease/condition and then any "other terms."2) To limit to open or soon to be open studies, select "Recruiting or and not yet recruiting studies." 3) To focus the list of studies (e.g., by age or country), use the filters on the left.
Link: https://clinicaltrials.gov/
Undiagnosed Diseases Network
Created by: NIH-funded research study
What is it? Research study that seeks to diagnose patients when healthcare providers are unable to discover a diagnosis for what is suspected to be a genetic condition.
Tips and Tricks: If a thorough medical evaluation has not resulted in a diagnosis, providers can provide a recommendation so patients can apply for inclusion in the study
Link: https://undiagnosed.hms.harvard.edu/
Genetics Resources Home
PREVIOUS: Support Families Resources 1/2
NEXT: Download Toolkit

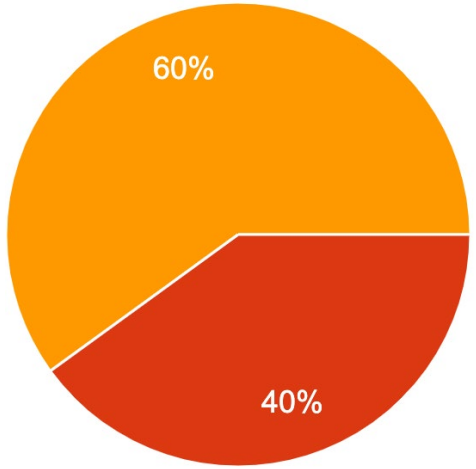
App Evaluation



What is your profession?

5 responses

- Nurse
- Physician Assistant
- Attending Physician
- Medical Resident or Fellow
- Neonatal Nurse Practitioner



How often do you interact with genetic testing for your career?

5 responses

- Daily
- Weekly
- Monthly
- Yearly
- Never or Almost Never

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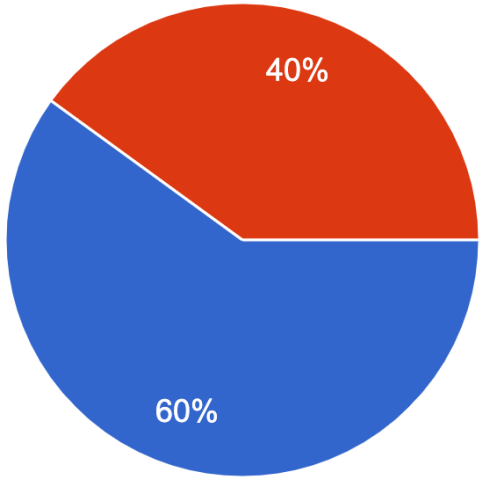
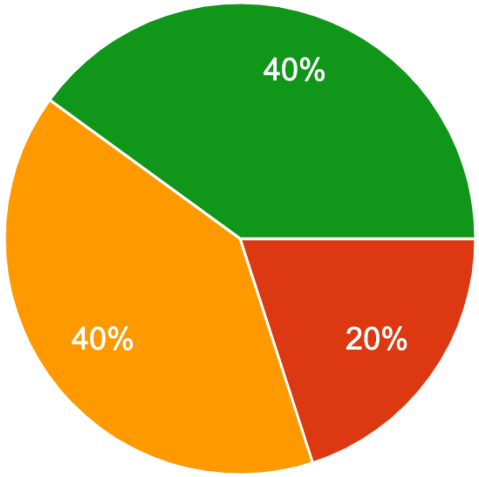
App Evaluation



What phone operating system (OS) did you use to access the app?

5 responses

- iPhone (iOS)
- Android



How would you describe your level of genetics knowledge?

5 responses

- Very High
- High
- Moderate
- Low
- Very Low

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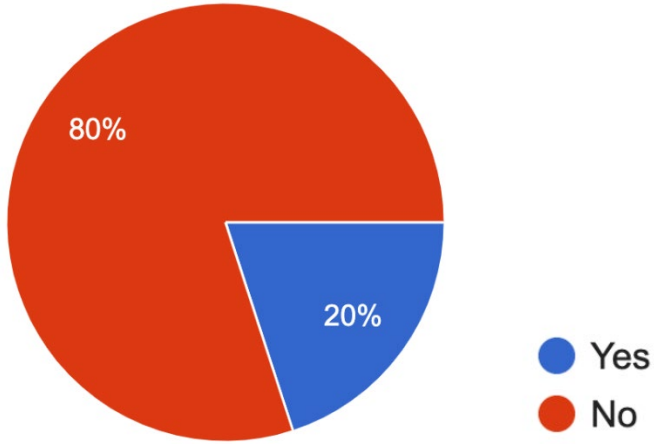
App Evaluation

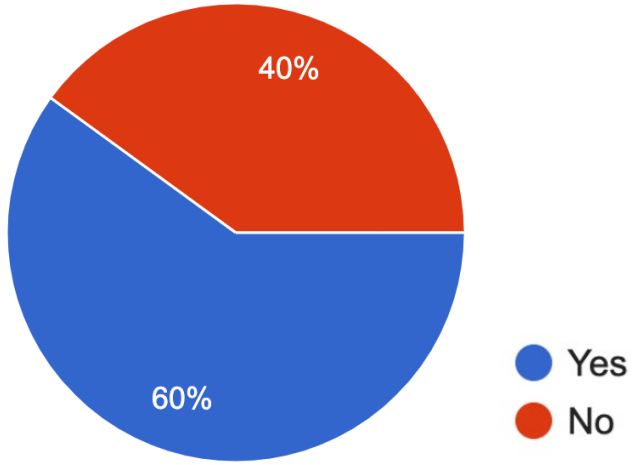
Did you need to access the additional instructions in order to utilize the app?

5 responses



Please add the app to your phone's home screen. Instructions for doing so can be accessed from the app home screen and once added, the app sho...ow. Were you able to add the app to your phone?

5 responses



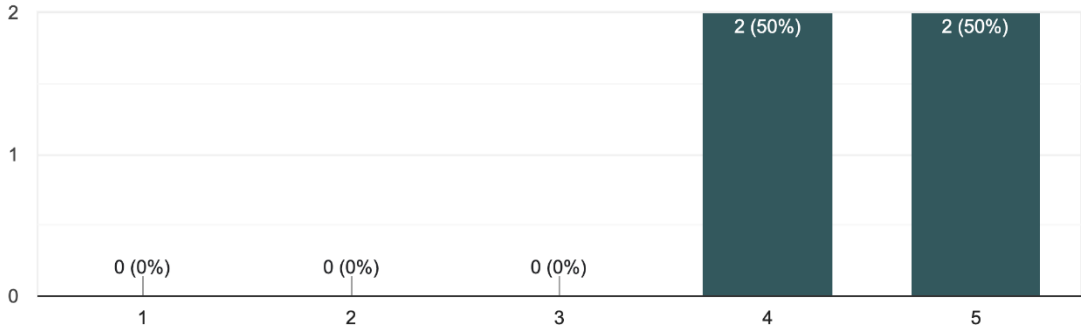
Inter-Society Coordinating Committee for Pra
PEG)

NIH
NHGRI

Evaluation

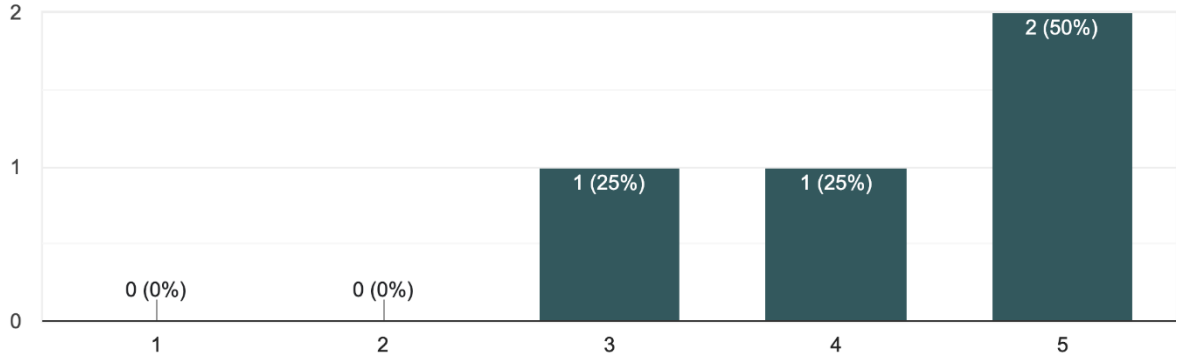
I imagine that most people would learn to use this app very quickly

4 responses



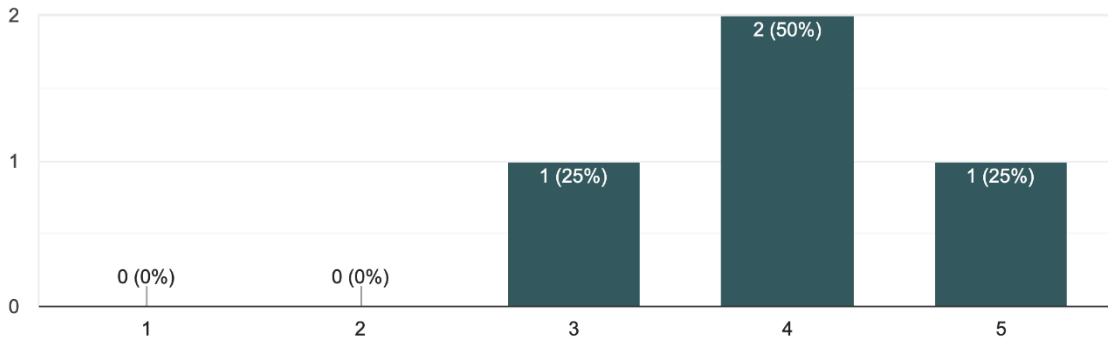
I think the app is easy to use

4 responses



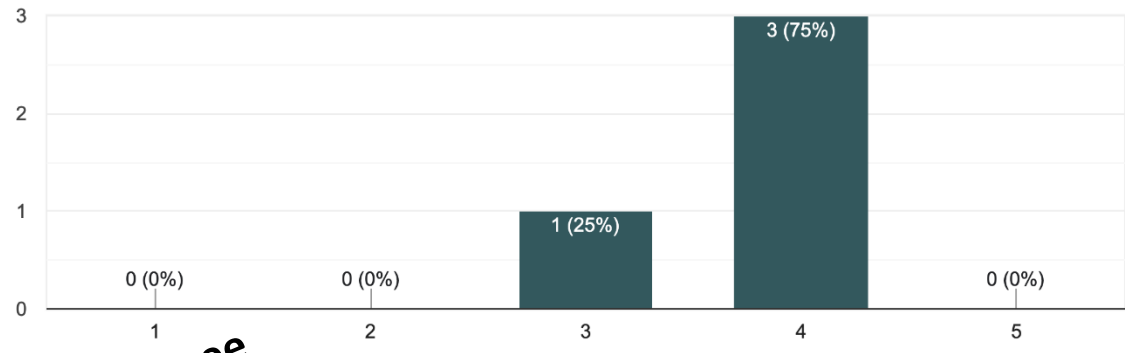
I think that I would like to use this app frequently

4 responses



I feel very confident using the app

4 responses



Strongly Disagree

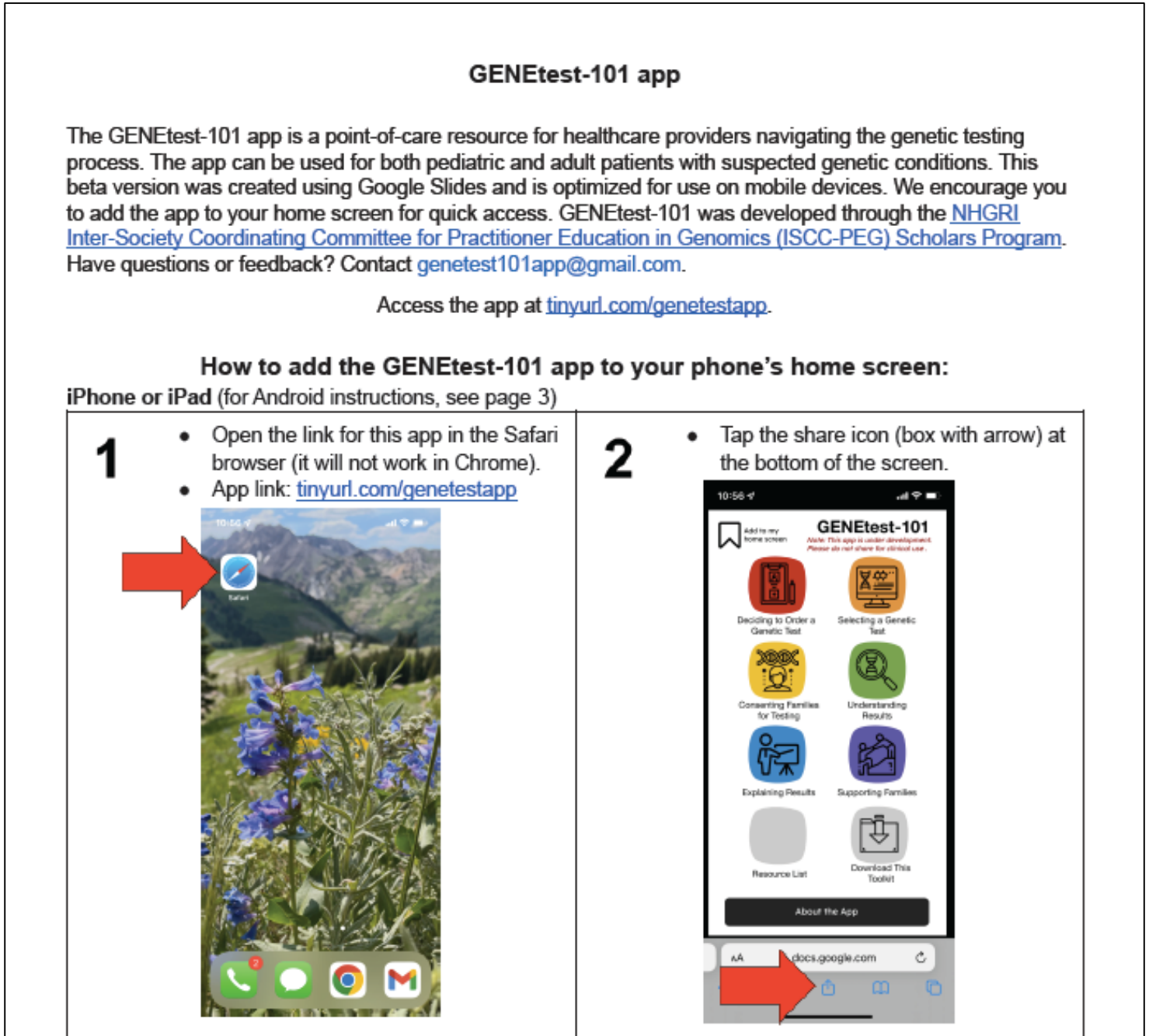
Strongly Agree

Evaluation

- Themes from open-ended questions:
  - Some acronyms and terminology may be unfamiliar to the audience
  - Standardize bullets and formatting
  - ”Home” and back buttons may not be intuitive for some
  - GENETEST-101 vs GENEtest-101
  - Easy to follow, good/many resources
  - Well-organized

App Revisions

- Pdf with app overview and instructions for adding it to home screen on iOS/Android
  - https://tinyurl.com/genetestapphome (pdf)
  - Also accessible from “Add to my home screen” button on app
- Made other suggested style and wording changes to increase consistency and accessibility



GENEtest-101 app

The GENEtest-101 app is a point-of-care resource for healthcare providers navigating the genetic testing process. The app can be used for both pediatric and adult patients with suspected genetic conditions. This beta version was created using Google Slides and is optimized for use on mobile devices. We encourage you to add the app to your home screen for quick access. GENEtest-101 was developed through the NHGRI Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG) Scholars Program. Have questions or feedback? Contact genetest101app@gmail.com.

Access the app at tinyurl.com/genetestapp.

How to add the GENEtest-101 app to your phone’s home screen:

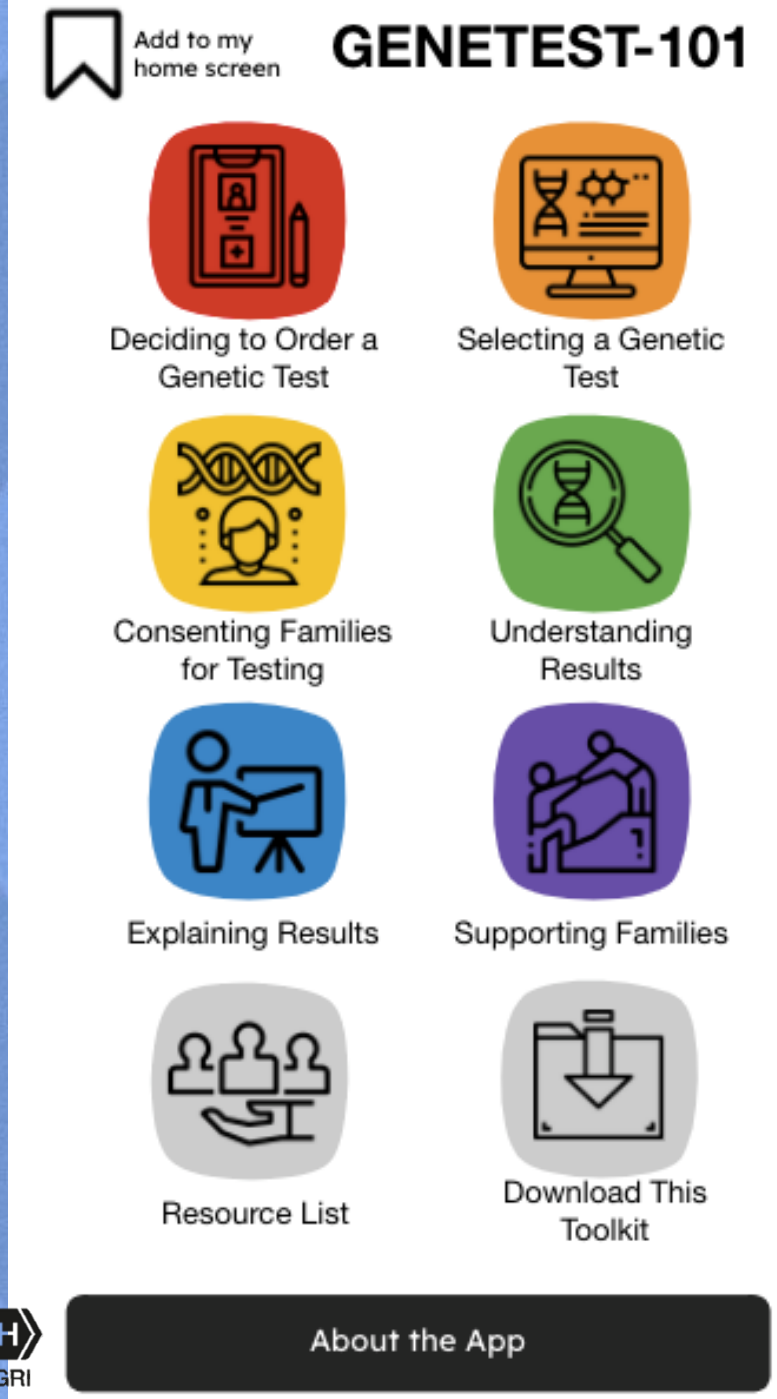
iPhone or iPad (for Android instructions, see page 3)

1

- Open the link for this app in the Safari browser (it will not work in Chrome).
- App link: tinyurl.com/genetestapp

2

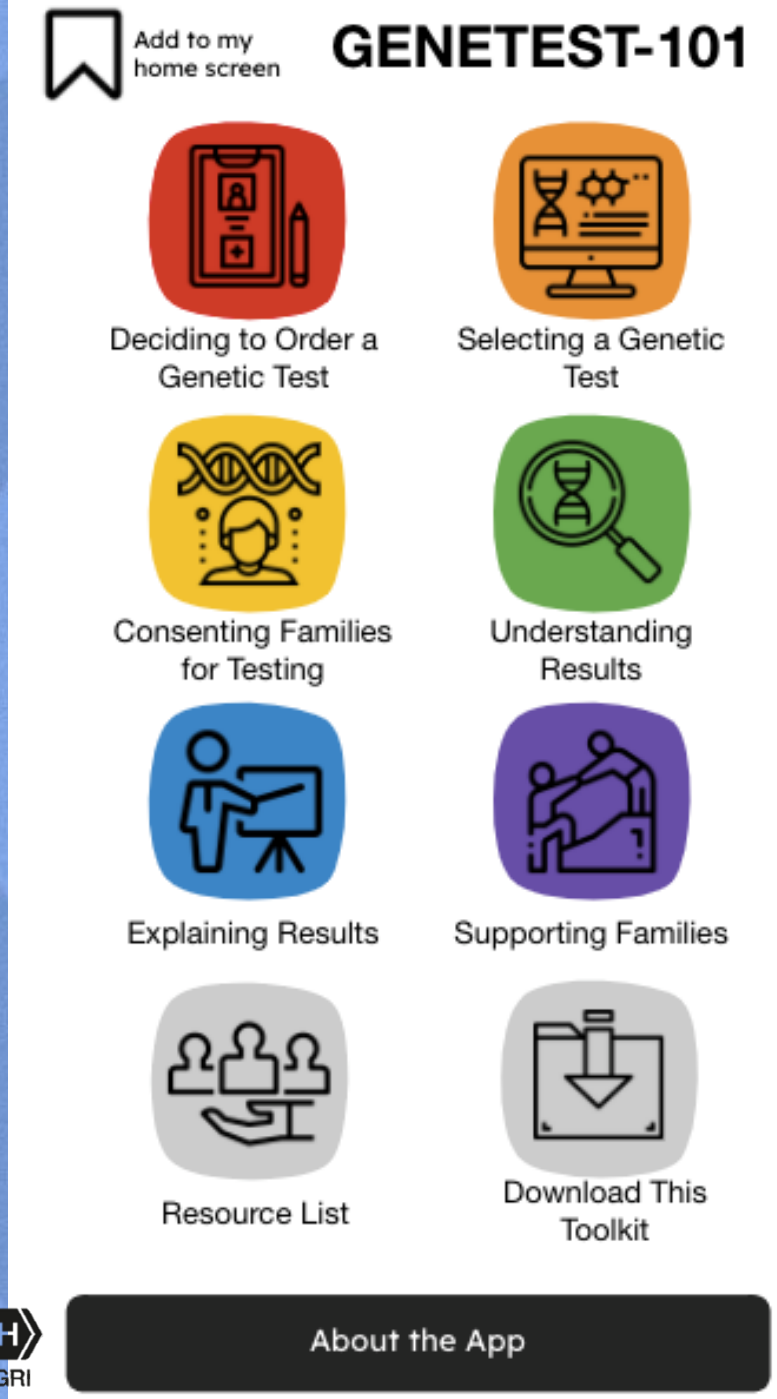
- Tap the share icon (box with arrow) at the bottom of the screen.



App Dissemination

- Submission to NHGRI's GenomeEd resource repository
- Sharing by ISCC-PEG membership
- Evaluation of other opportunities to share and improve app prototype
- Suggestions or opportunities to share are welcomed!

Scan to access app



Questions?

Thank you to my mentor Michelle Snyder, ISCC-PEG Rare Disease Project Group members including Dr. Sabrina Malone-Jenkins, Rachel Palmquist, Kristen Fishler, Dr. Leah Burke, Janine Lewis, and ISCC-PEG Co-chairs Dr. Rich Haspel and Dr. Donna Messersmith for all their help and guidance throughout this project.

App Instructions:
tinyurl.com/genetestapphome
App link: tinyurl.com/genetestapp
Questions or feedback? Contact genetest101app@gmail.com.

Scan to access app