



Understanding Genetics Learning Needs of People Affected by Rare Disease

Laura Quinn¹, Kendall Davis², Ashley Yee², Karen Leydiker³, Holly Snyder⁴

1. Assai Health Solutions 2. Global Genes 3. Invitae 4. Illumina

Background

Global Genes (GG) is a non-profit organization that works to educate, equip and empower the global rare disease (RD) community to help eliminate the challenges of RD. The GG *RARE Foundation Alliance* is a coalition of over 500 RD organizations. *RareUniversity* is a GG online learning initiative that aims to provide free education on topics salient for people affected by RD.

GG approached Assai Health Solutions to develop a genetics course specifically tailored to the needs of people affected by RD. Development began with learner needs research to inform the scope, content and features of the course.

Objectives

To understand desired content, features, and customization of an online genetics course for learners who are directly or indirectly affected by RD.

Methods

12-question online-survey (n=586)
Emailed to 586 GG RARE Foundation Alliance members*

Responses analyzed and used to develop focus groups (FG).

Two FGs, 120 minutes each (n=5, n=3)

Survey

- Quantitative questions (n=10) analyzed using SurveyPlanet™ program.
- Responses to qualitative questions (n=2) analyzed using standard coding methodology.

Focus Groups (FG)

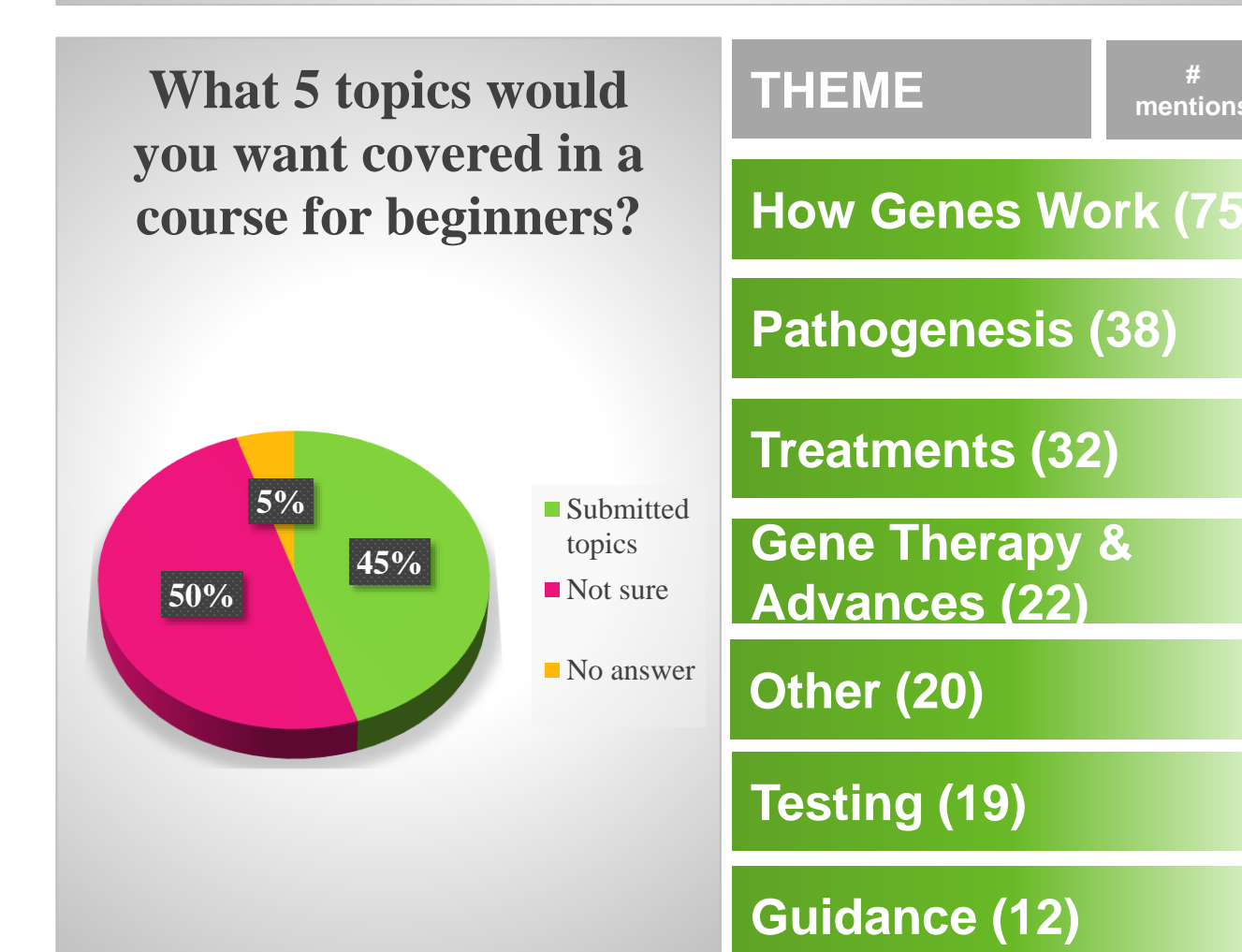
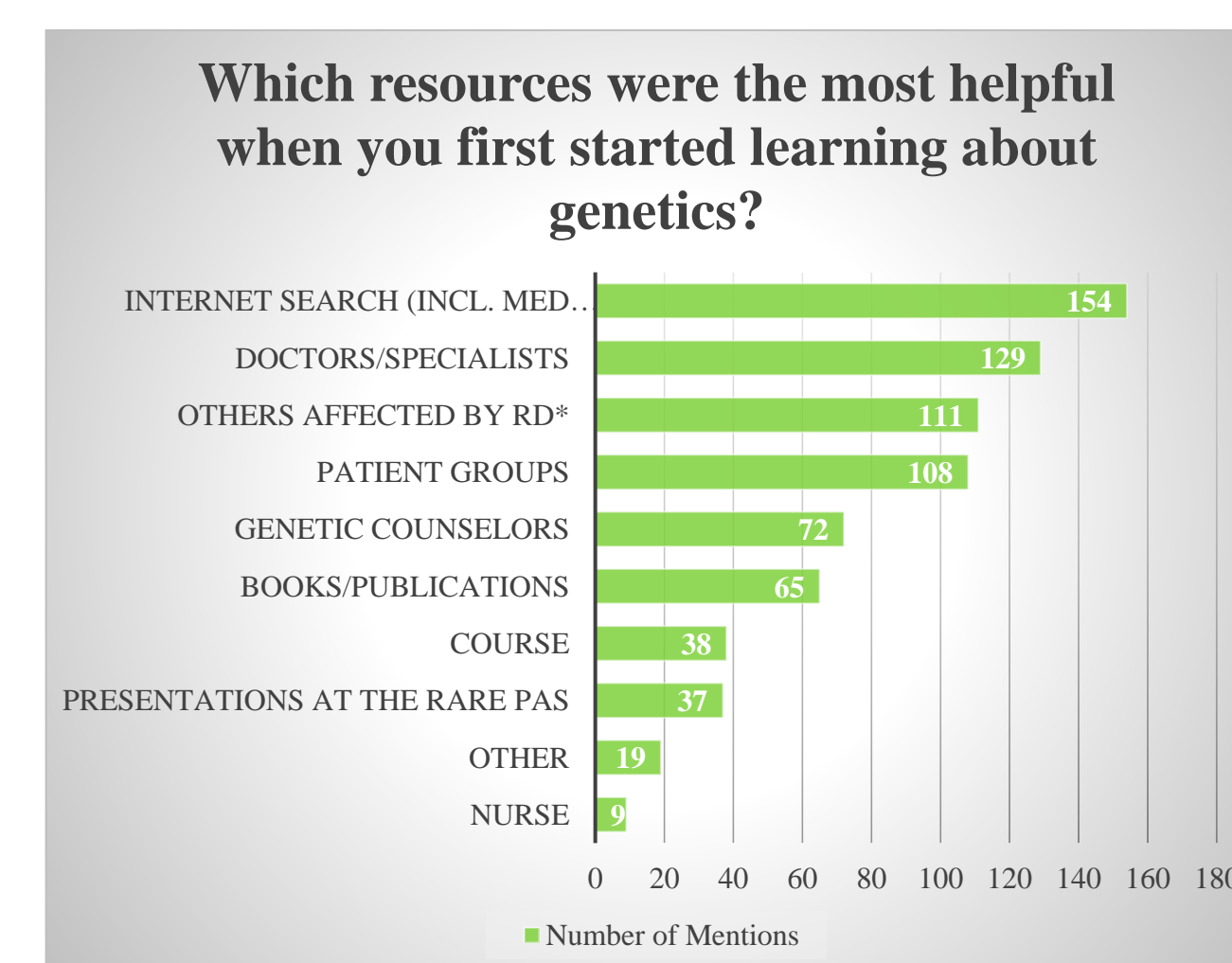
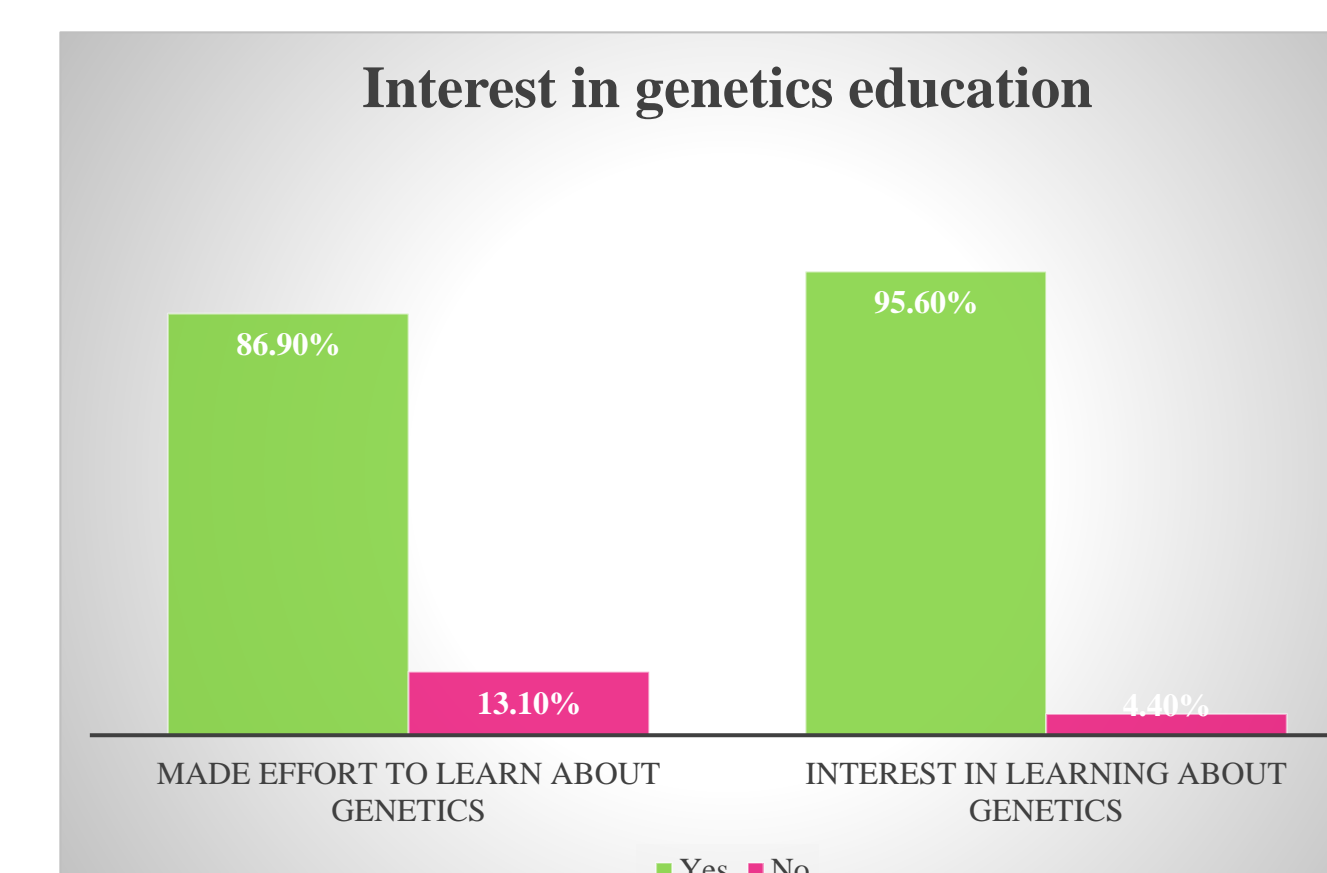
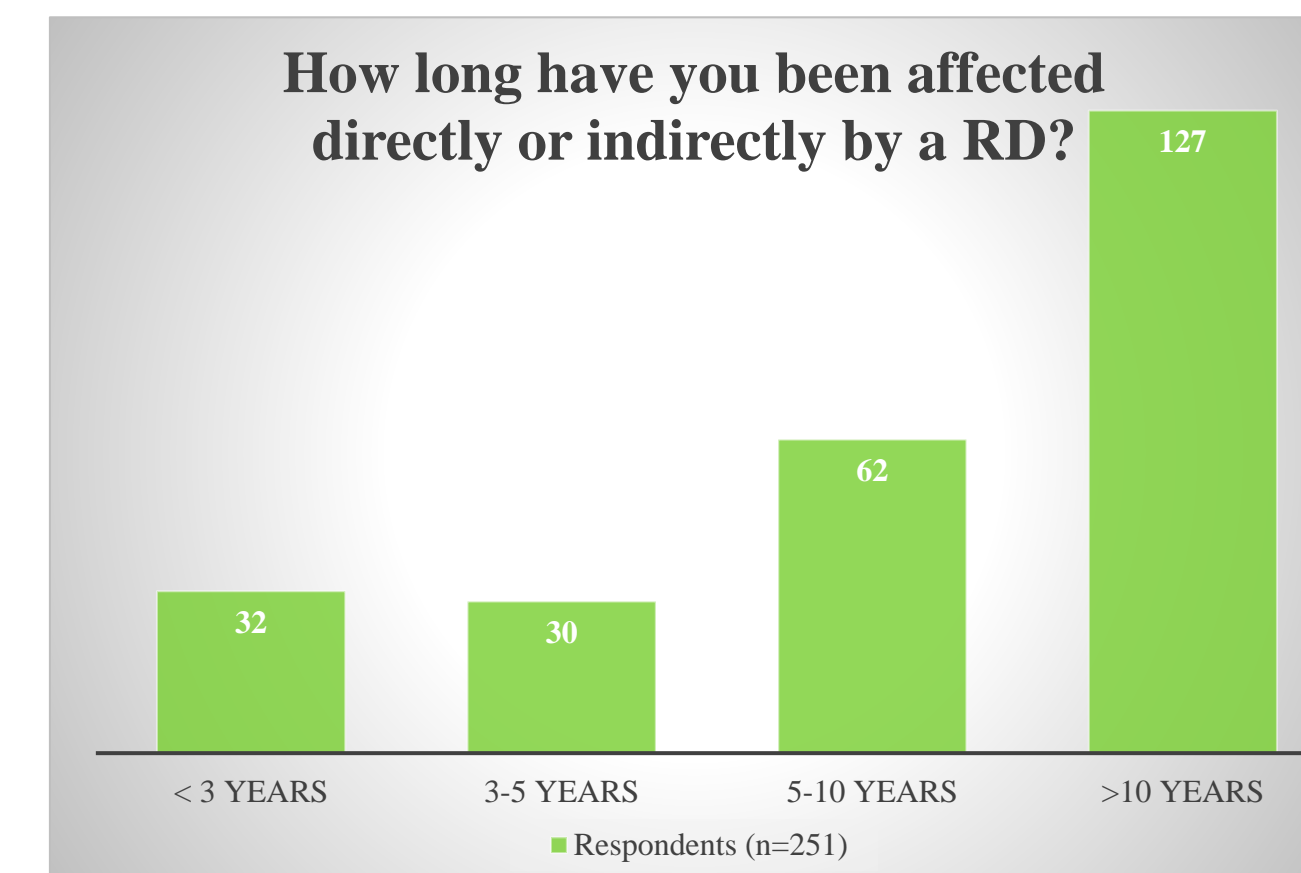
- Participants selected to represent a range of profiles of people affected by RD.
- Explored learner expectations for course structure and customization.
- Conducted via video conference. Discussions were video recorded, transcribed and analyzed.

* Request made for organizations to forward survey on to their own network

Limitations

Online survey respondents mostly resident in US (90%) and had significant RD experience (87% > 3 years of experience); Small focus group sample size; All FG participants were affiliated with RARE Foundation Alliance member organizations and US residents; Coding and analysis for both online survey and FG output conducted by same researcher; FG analysis not validated with participants due to capacity constraints

Results



Learner Expectations: Learning Outcomes

BE	DO	FEEL
<ul style="list-style-type: none"> Aware of and able to apply genetic concepts Able to connect the genetics concepts to own circumstances 	<ul style="list-style-type: none"> Communicate effectively with health care providers Make important decisions regarding treatments and research for themselves or loved ones Communicate clearly to entire ecosystem (i.e. family and friends) Make situation appreciable to other stakeholders (e.g. child's school, TSA, etc.) 	<ul style="list-style-type: none"> Confident in navigating interactions Reassured they are making the best choices and acting on accurate information

Learner Expectations: Course Scope, Content, Features

Scope	<ul style="list-style-type: none"> Gene/genome function Pathogenesis Management Gene therapy & scientific advances Genetic testing
Duration	6 – 12 weeks
Pacing	<ul style="list-style-type: none"> Self-paced, average of 1-2 hour of learning per week No pre-determined learner journey (choose own adventure)
Format	<ul style="list-style-type: none"> Easily referenced as needed over time Mix of video, audio, text catering to multiple learning styles Easy-to-navigate platform
Content	<ul style="list-style-type: none"> Suitable for individuals with varying levels of knowledge Explanation of genetic concepts relevant to RD beyond 'basic' genetics concepts Ready-to-use content to support navigating important interactions Medically accurate and peer-reviewed
Tone/Style	<ul style="list-style-type: none"> Simple language, especially to explain complex genetic concepts Empathetic tone Considerate of cultural diversity Provides hope or does not take away hope
Interactivity	<ul style="list-style-type: none"> Opportunity to ask questions Availability of a mentor/administrator Enables sharing of information
Special considerations	<ul style="list-style-type: none"> Negative outcomes, heredity and comorbidity discussed with special consideration for learner frame of mind Does not describe features/experiences/outcomes as universally representative Parental sign-off for learners under age 18
Duty of care	<ul style="list-style-type: none"> Non-prescriptive content Information and tools to support decision making Efforts made to provide robust sign-posting rather than relying on 'talk to your doctor'

Conclusion

Research identified high levels of interest in genetics education for individuals across all stages of the RD journey. Desired learning outcomes and course features enabled development of a comprehensive genetics course.

- Genetic concepts are explored in 4 contexts: *Genetics & RD, Family & Heredity, Genetic Testing, Scientific Advances*.
- Special focus on 'practical applicability' of genetic concepts and development of skills to apply concepts (i.e. online research skills).
- Content/structure allows learners to start with topics of most interest to them.
- Due to budget and capacity constraints the course scope does not currently include access to a mentor or administrator.

Implications

- Significant interest in genetics education exists for both highly experienced RD patients and newcomers (one possible reason for this may be the pace of advances in genetics).
- Potentially significant need for skills-development/ practical application support to accompany genetics education. This may increase confidence and reduce anxiety.
- Although interactive learning with a health care professional is generally preferred, there is significant interest in online learning resources.

Further Research

- Survey users to evaluate course scope, format and customization.
- Research barriers exist in locating/ accessing online resources
- Assess which forms of customized learning are most effective for users.
- Follow up survey with this cohort to assess if course met learning needs.
- Survey additional advocacy groups to determine effectiveness of curriculum and generalizability to broader community.
- Survey additional groups such as individuals seeking genetic testing for multifactorial conditions to determine if curriculum is valuable beyond RD.