



# Understanding Genetics Learning Needs of People Affected by Rare Disease

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## 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)

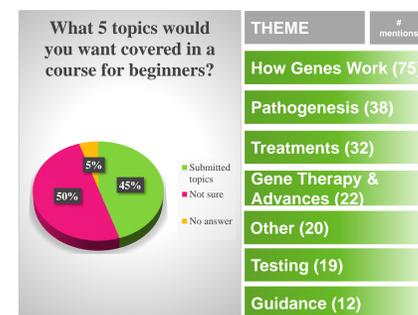
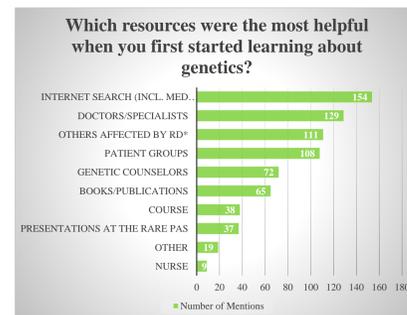
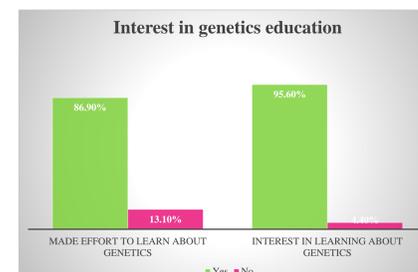
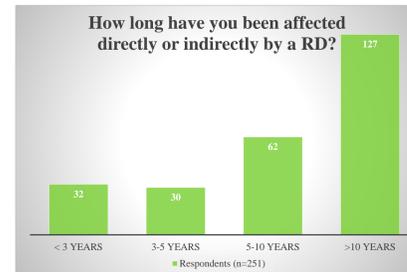
- Participants selected to represent a range of profiles of people affected by RD.
- Explored learner expectations for course structure and customization.
- Conducted via video conference. Discussions were video recorded, transcribed and analyzed.

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

## Learner Expectations: Course Scope, Content, Features

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

## 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.