

ISCC Rare Disease Project Group Update

Michelle Snyder
February 25, 2020



ISCC Rare Disease Project Group

- Goals

- Educate healthcare professionals on available resources for rare genetic diseases.
- Develop resources to address the challenges of rare diseases, such as diagnostic delays, lack of available treatment guidelines, and limited referral pathways.
- Identify effective dissemination methods for information about rare diseases by specialty or practice area.



National Center
for Advancing
Translational Sciences

Genomic Literacy Survey Question

- When diagnosing and/or managing patients with rare diseases or conditions, in which of the following areas have you encountered difficulties? (select as many as apply)
 - Lack of diagnostic guidelines
 - Lack of access to diagnostic tests
 - Delay in, or inability to make a definitive diagnosis
 - Lack of treatment/management guidelines
 - Lack of available treatments
 - Uncertainty about where to refer to
 - Difficulties in accessing specialized clinics/services
 - Difficulties in accessing allied health services (e.g. physio, speech, OT, psychology etc.)
 - Difficulties accessing genetic testing/services
 - Uncertainty about available peer support groups for the patient and his/her family



What dissemination method would be best?

- Repeated vs. one-time
- Mobile accessible**
- Shareable
- Reusable/repurpose-able
- Conversational language
- Organization buy-in
- Accessible metrics (web analytics, shares, etc.)



Areas of Interest

- Trainee education modules
- Slide deck
- Podcast
- Social media

Working group session:

Matrix of important rare disease resources/topics

<https://app.mural.co/t/hs28383/m/hs28383/1582649350879/7692c2a0abec08b47c3823302091e43e06838b49>



National Center
for Advancing
Translational Sciences

Asks

- Any organizations interested in piloting trainee education on rare diseases?
- What type of content dissemination is most successful for your organization (webinars, newsletters, blogs, social media, articles, etc.)?



National Center
for Advancing
Translational Sciences