ISCC Rare Disease Project Group Update

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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.
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
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.)?