“If not me, who?”

Awareness, Stigma, and Advocacy Experiences Among Adults With Rare Disorders

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Rare Diseases, Disorders, or Disabilities (RDs)

- Affects < 200,000 people in U.S. = RD
  
  (Rare Diseases Act of 2002, & Orphan Drug Act of 1983 (Section 526))

- ≈ 7,000 different RDs
  - Affect 1 in 15 people worldwide (de Vrueh et al., 2013)

- RDs are Individually Rare, but Collectively Pervasive
Lack of RD Awareness → Common Challenges*

**Diagnostic Odysseys**
- It is difficult and time-consuming to get correctly diagnosed with an RD.
  
  (Black et al., 2015; EURORDIS, 2007)

**Lack of Treatments**
- Only 5% of RDs have any FDA-approved treatment option.

  (H. Res. 1154, 2018)

**Stigma**
- Judged, socially devalued, excluded, marked as other or different.
  
  (Bogart et al., 2018; Kurzban & Leary, 2001; Zhu et al., 2017)
- Enacted stigma → anticipated & internalized stigma → negative impacts on quality of life
  
  (Earnshaw & Quinn, 2012)

* (Bogart et al., 2011; Bogart, 2015; Bogart et al., 2018; Kurzban & Leary, 2001; Orphanet, 2019; Zhu et al., 2017)
Study 1: Two Focus Groups (WebEx)

Participants

- Pool = AWaRDS Phase 1 participants
- All had/have RDs
- 9 total
- 5 Male, 4 Female
- 7 “White,” 2 “Black or African American”
- Median household income: $30,001 - $45,000 per year
## Study 2: Eighteen Advocate Interviews (Zoom)

### Demographic Characteristics of RD Advocate Interviewees

<table>
<thead>
<tr>
<th>Demographic Characteristic</th>
<th>Aggregate Data</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age</strong></td>
<td><em>Age Range</em>: 21-73 years</td>
</tr>
<tr>
<td></td>
<td><em>Mean Age</em>: 42.61 years</td>
</tr>
<tr>
<td><strong>Gender Identification</strong></td>
<td><em>Female</em>: 10</td>
</tr>
<tr>
<td></td>
<td><em>Male</em>: 6</td>
</tr>
<tr>
<td></td>
<td><em>Other</em>: 2</td>
</tr>
<tr>
<td><strong>Ethnicity and/or Race</strong></td>
<td><em>Black or African American</em>: 2</td>
</tr>
<tr>
<td></td>
<td><em>White / Caucasian</em>: 12</td>
</tr>
<tr>
<td></td>
<td>*Identified as Multiple Categories or “Other”: 4</td>
</tr>
<tr>
<td><strong>Household Income Bracket</strong></td>
<td><em>Median</em>: $45,000-$60,000</td>
</tr>
<tr>
<td></td>
<td><em>Mode</em>: $90,000 and above</td>
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</tbody>
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## RDs Represented by Participants in Studies 1 & 2

<table>
<thead>
<tr>
<th>Study 1: Focus Groups</th>
<th>Study 2: Advocate Interviews</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>RDs</strong></td>
<td><strong>RDs</strong></td>
</tr>
<tr>
<td>Friedrich’s Ataxia</td>
<td>Arteriovenous Malformation (AVM)</td>
</tr>
<tr>
<td>Klippel-Feil Syndrome, Type II</td>
<td>Bronchiectasis</td>
</tr>
<tr>
<td>Lymphomatoid Papulosis</td>
<td>Cauda Equina Syndrome (CES)</td>
</tr>
<tr>
<td>Membranous Nephropathy</td>
<td>Cervical Dystonia</td>
</tr>
<tr>
<td>Moebius Syndrome</td>
<td>Charcot-Marie-Tooth Disease (CMT), type 2A2</td>
</tr>
<tr>
<td>Myopic Macular Degeneration</td>
<td>Common Variable Immune Deficiency (CVID)</td>
</tr>
<tr>
<td>Spinocerebellar Ataxia (non-specific)</td>
<td>Congenital Vocal Cord Paralysis (Congenital Laryngeal Palsy)</td>
</tr>
<tr>
<td>Spinocerebellar Ataxia (SCA) 8</td>
<td>Crouzon Syndrome</td>
</tr>
<tr>
<td>Spinocerebellar Ataxia (SCA), type 2</td>
<td>Diabetes Insipidus (DI)</td>
</tr>
<tr>
<td></td>
<td>Ehlers-Danlos Syndrome (EDS)</td>
</tr>
<tr>
<td></td>
<td>Enoyl-CoA Hydratase 1 Deficiency (Deficiency of Gene ECHS1)</td>
</tr>
<tr>
<td>Goldenhar Syndrome</td>
<td>Familial or Familiar Hemiplegic Migraines (FHM)</td>
</tr>
<tr>
<td>Guillain-Barré Syndrome (GBS), subcategory: acute motor axonal neuropathy</td>
<td>Fanconi Anemia (FA)</td>
</tr>
<tr>
<td>Idiopathic hypersomnia (IH)</td>
<td>Goldenhar Syndrome</td>
</tr>
<tr>
<td>Inclusion Body Myopathy, with early-onset Paget disease of bone, associated with Frontotemporal Dementia (IBM-PFD) [a.k.a Valosin Containing Protein (VCP) Disease] [a.k.a. Multisystem Proteinopathy (MSP1)]</td>
<td>Langerhans Cell Histiocytosis (LCH)</td>
</tr>
<tr>
<td>Idiopathic hypersomnia (IH)</td>
<td>Leiomyosarcoma (LMS)</td>
</tr>
<tr>
<td>Narcolepsy, type 2 (no cataplexy)</td>
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</tr>
<tr>
<td>Osteonecrosis (rare form)</td>
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</tr>
<tr>
<td>Pigmented Villonodular Synovitis (PVNS)</td>
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</tr>
<tr>
<td>Spinocerebellar Ataxia (SCA), type 2</td>
<td>Spinocerebellar Ataxia (SCA), type 2</td>
</tr>
<tr>
<td>Sutton’s Disease (Sutton Disease 2)</td>
<td>Sutton’s Disease (Sutton Disease 2)</td>
</tr>
<tr>
<td>Triple X syndrome</td>
<td>Triple X syndrome</td>
</tr>
<tr>
<td>Undiagnosed adult-onset RD: rare symptom cluster, including severe chronic pain and auto-immune chronic urticaria</td>
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<tr>
<td>VATER (sometimes known as VACTERL) Syndrome</td>
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Data Analysis

• Thematic Coding & Analysis
  ❖ Transcripts → Codes → Themes

• Focused on...
  ❖ Study 1: statements involving RD awareness, advocacy, & experienced stigma
  ❖ Study 2: statements involving advocacy, and recommendations for increasing RD awareness and/or decreasing RD stigma
Objectives

From People with RDs...

- Gather Statements about Awareness, Stigma, and Advocacy Experiences
- Get Concrete Suggestions about Increasing RD Awareness and Decreasing RD Stigma
Study 1: Focus Groups

- Interpersonal
- Global/Social

Expression of Disability
- Avoidant
- Chosen
- Not Chosen

Working Towards Awareness
- Medical

Experience of Stigma
- Personal
- Medical
...the social aspects of the conditions are probably the worst. People’s first impressions are sometimes incorrect. And sometimes I can get past that, and sometimes I can’t.
Study 2: Advocate Interviews

Recommendations for Individuals & Organizations
- Conduct Interpersonal Education
- Work for Systemic Change
- Recommendations For Researchers

Societal & Structural Roadblocks
- Discomfort & Invalidation
- Limitations of Time, Energy, & Reality
Concrete Ideas for Increasing Awareness

• **Individual Level:**
  ▪ Be prepared to be an advocate.
  ▪ Tell your story and amplify others’.

• **Larger Scale:**
  ▪ More Representation
  ▪ Lobby Policymakers for...
    ▪ More Funding, Research
    ▪ More Inclusive and Equitable Social Supports
  ▪ Educate medical professionals, students, children, and others.

  ➢ *Key: Use the voices of people with RDs*
It’s important to tell people about your condition, whenever you can, because they don’t know.
...awareness among doctors... is very low. And so, my thought would be to start with the doctors. Would be start with medical school students.... and let them know that we're not just these rarities in these textbooks. That we exist as people and we're not just here to be your patient, we're trying to be people.
Study 2: Advocate Interviews

Recommendations for Individuals & Organizations
- Conduct Interpersonal Education
- Work for Systemic Change
- Recommendations For Researchers

Societal & Structural Roadblocks
- Discomfort & Invalidation
- Limitations of Time, Energy, & Reality
People with disabilities… we have the least rights of anybody… *because* we can't protest en masse, to even *have* awareness…. They make life *so hard* to be out of the house. It’s like… “We know you can’t really fight us.”
What Did We Learn?
Future Directions

→ I will...
  • Be an advocate.
  • Keep gathering data, input from people with RDs.

→ As I...
  • Construct applied awareness and stigma interventions.
  • Test interventions’ effectiveness in the real world.
After somebody passes away, if they've been an advocate, they still go forward. And, somebody who has not advocated for it, and has just lived with it, does not. So, it is part of a legacy.... Some of the things of what I've been doing... it's not all for naught.... It's something that... that will go beyond me. And advocacy does that.
References


EURORDIS: Rare Diseases Europe. (2007). Survey of the delay in diagnosis for 8 rare diseases in Europe (‘EURORDIS Care 2’). https://www.eurordis.org/content/undiagnosed-rare-diseases


THANK YOU!