

“If not me, who?”

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

Emily F. Plackowski & Kathleen R. Bogart



**Oregon State
University**

AWaRDS Study
Adults with **Rare Disorders** Support Study





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Background

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))

- \approx 7,000 different RDs
 - Affect 1 in 15 people worldwide (de Vruet 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

Demographic Characteristic	Aggregate Data
Age	Age Range: 21-73 years Mean Age: 42.61 years
Gender Identification	Female: 10 Male: 6 Other: 2
Ethnicity and/or Race	Black or African American: 2 White / Caucasian: 12 Identified as Multiple Categories or "Other": 4
Household Income Bracket	Median: \$45,000-\$60,000 Mode: \$90,000 and above



RDs Represented by Participants in Studies 1 & 2

Study 1: Focus Groups

Friedrich's Ataxia
Klippel-Feil Syndrome, Type II
Lymphomatoid Papulosis
Membranous Nephropathy
Moebius Syndrome
Myopic Macular Degeneration
Spinocerebellar Ataxia (non-specific)
Spinocerebellar Ataxia (SCA) 8
Spinocerebellar Ataxia (SCA), type 2

Study 2: Advocate Interviews

Arteriovenous Malformation (AVM)
Bronchiectasis
Cauda Equina Syndrome (CES)
Cervical Dystonia
Charcot-Marie-Tooth Disease (CMT), type 2A2
Common Variable Immune Deficiency (CVID)
Congenital Vocal Cord Paralysis (Congenital Laryngeal Palsy)
Crouzon Syndrome
Diabetes Insipidus (DI)
Ehlers-Danlos Syndrome (EDS)
Enoyl-CoA Hydratase 1 Deficiency (Deficiency of Gene ECHS1)
Familial or Familial Hemiplegic Migraines (FHM)
Fanconi Anemia (FA)
Goldenhar Syndrome
Guillain-Barré Syndrome (GBS), subcategory: acute motor axonal neuropathy
Idiopathic hypersomnia (IH)
Inclusion Body Myopathy, with early-onset Paget disease of bone, associated with Frontotemporal Dementia (IBMPFD) [a.k.a Valosin Containing Protein (VCP) Disease] [a.k.a. Multisystem Proteinopathy (MSP1)]
Langerhans Cell Histiocytosis (LCH)
Leiomyosarcoma (LMS)
Mast Cell Activation Syndrome (MCAS) [a.k.a. Mast Cell Response]
Narcolepsy, type 2 (no cataplexy)
Osteonecrosis (rare form)
Pigmented Villonodular Synovitis (PVNS)
Spinocerebellar Ataxia (SCA), type 2
Sutton's Disease (Sutton Disease 2)
Triple X syndrome
Undiagnosed adult-onset RD: rare symptom cluster, including severe chronic pain and auto-immune chronic urticaria
VATER (sometimes known as VACTERL) Syndrome



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



An abstract graphic design featuring a dark gray background. On the left, a ribbon with a black and white zebra stripe pattern is tied into a knot. A large, semi-transparent white circle is positioned behind the knot. To the right of the circle, the word "Results" is written in a white, serif font.

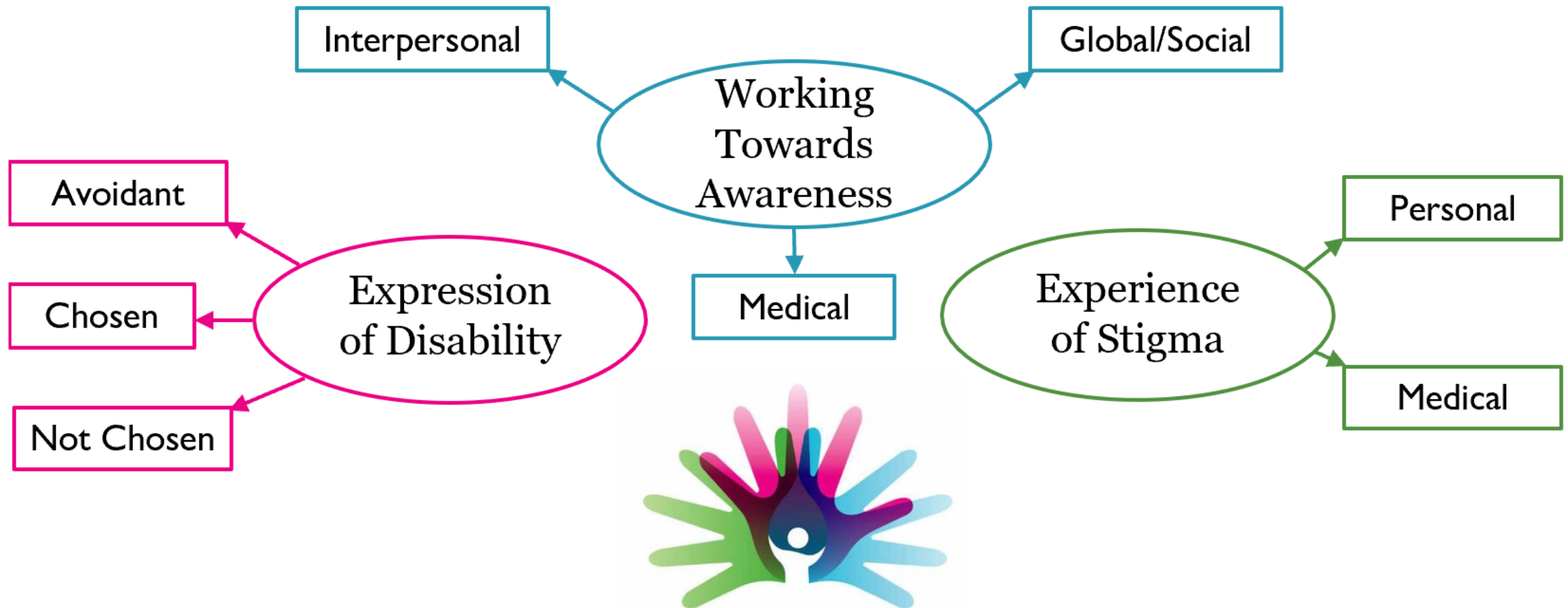
Results



**Themes
+
Sub-themes**

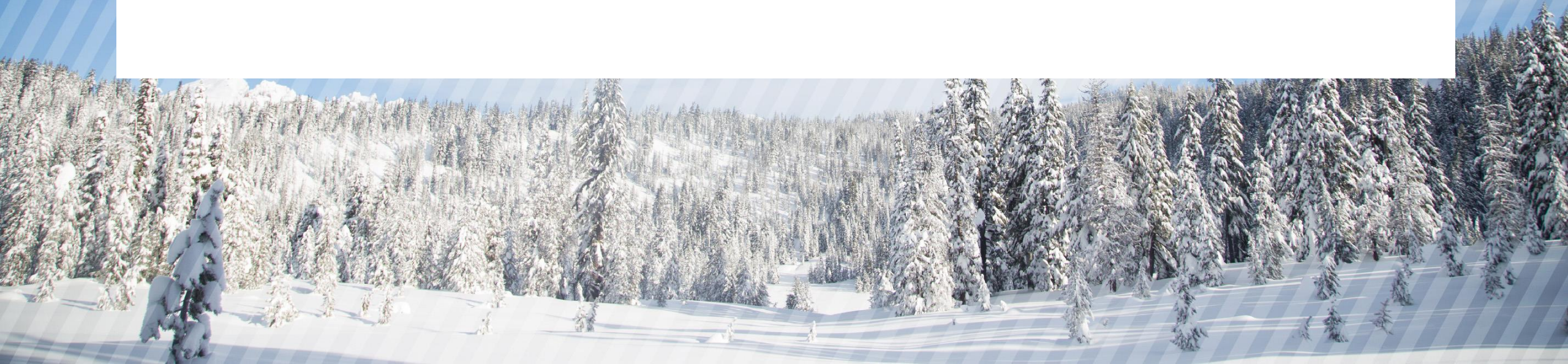


Study 1: Focus Groups



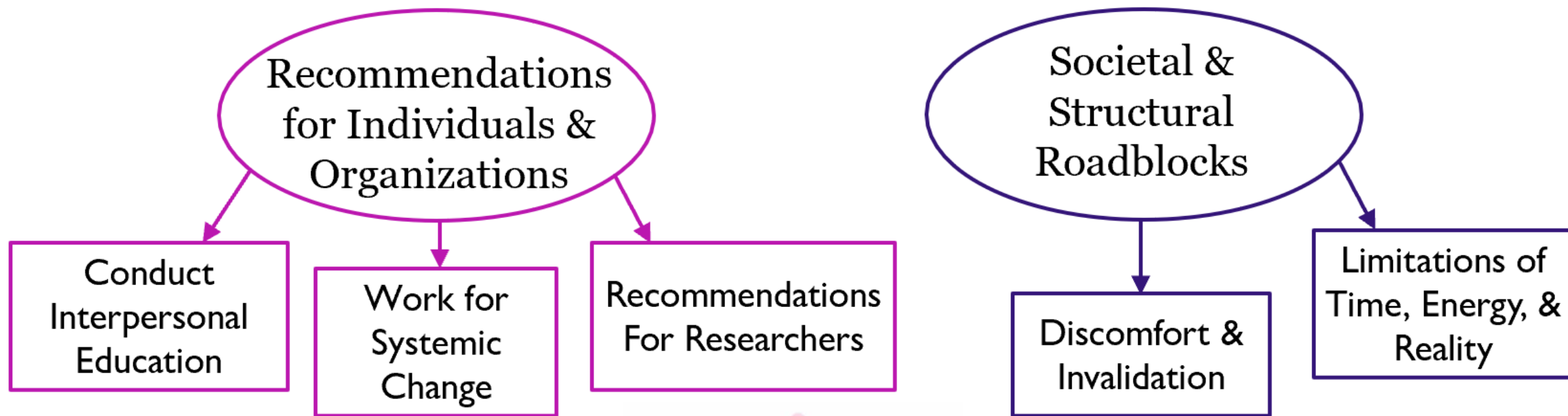


...the social aspects of the conditions are probably the worst. People's first impressions are sometimes incorrect. And sometimes I can get past that, and sometimes I can't.





Study 2: Advocate Interviews



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.



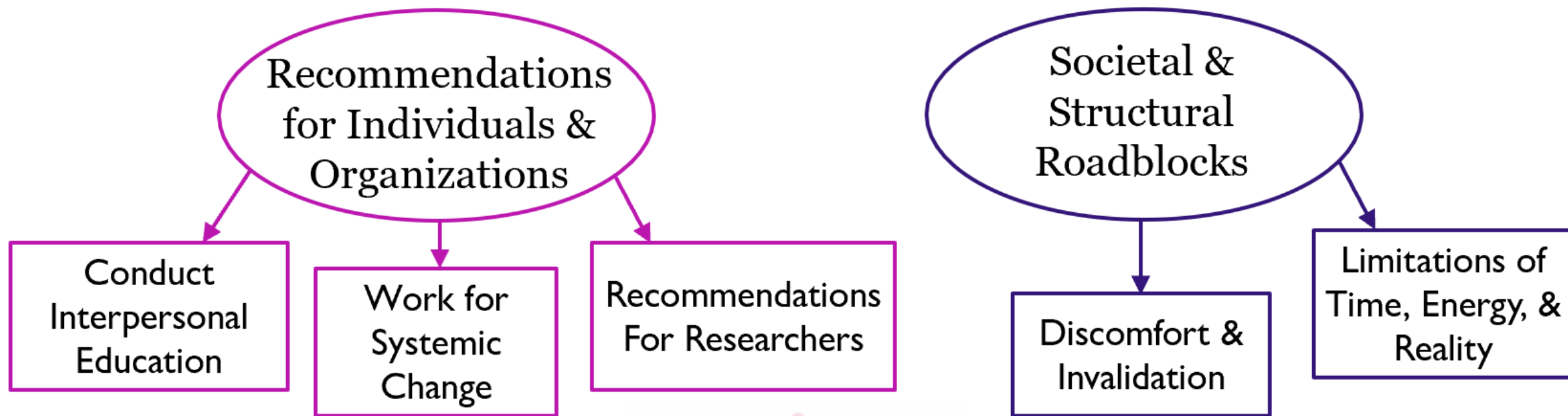


...awareness among doctors... is very low. And so, my thought would be to *start* with the doctors. Would be *start* with medical school students.... and let them know that we're not just these rarities in these textbooks. That we exist as people and we're not just here to be your patient, we're trying to be people.





Study 2: Advocate Interviews





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.



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THANK YOU!



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