Supporting those with rare disease

An examination of social support among adults from the United States with Sickle Cell Disease, Alopecia Areata, and Idiopathic Hypersomnia

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Background

- Rare diseases are those that affect a small number of people in the general population, but the collective burden of rare disease is considerable.[1]
- In the United States, greater than 25 million people are affected by a rare disease, and most of these diseases are without approved therapies from the Federal Drug Administration.[2]
- Due to the lack of approved therapies, social support (SS) may be an essential resource for those living with rare disease, but access to SS may vary by disease.

Objective

To describe and compare the distribution of SS among patients with sickle cell disease, alopecia areata, and idiopathic hypersomnia.

Sickle Cell Disease (SCD): A rare blood disorder that is inherited in an autosomal recessive manner. It is characterized by the presence of sickle, or crescent-shaped, red blood cells (erythrocytes) in the bloodstream.[3]

Idiopathic Hypersomnia (IH): A neurological sleep disorder that can affect many aspects of a person's life. Symptoms often begin between adolescence and young adulthood and develop over weeks to months. People with idiopathic hypersomnia have a hard time staying awake and alert during the day (chronic excessive daytime sleepiness, or EDS).[6]

Alopecia Areata (AA): A disorder characterized by loss of hair. Sometimes, this means simply a few bare patches on the scalp. In other cases, hair loss is more extensive.[5]

Methods

Data source and sample

- A cross-sectional analysis was conducted using data from the 2022 United States National Health and Wellness Survey (NHWS). The NHWS is, an online patient-reported outcomes survey administered to adults and is designed to be representative of the general population, relative to age, sex, and race/ethnicity.[8]
- We included adults who self-reported a physician diagnosis for SCD, AA, or IH

Measures

- Sociodemographic and health characteristics were used to describe the individual disease cohorts
- SS was measured using the Modified Medical outcomes Study Social Support Survey (mMOS-SS)[9]
  - Three summary scores were derived:
    1. Overall Support: emotional + instrumental support
    2. Emotional Support: "if you needed it, how often is someone available who understands your problems?"
    3. Instrumental Support: "If you needed it, how often is someone available to take you to the doctor?"
- To aid interpretation, summary scores were transformed to a scale of 0 to 100, with a score of 50 representing the average score for the full analytic sample

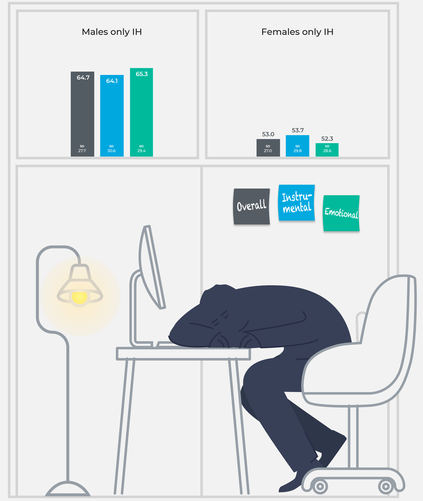
Analytic approach

- Levels of SS (all types) were examined within each disease cohort, stratified by gender, using descriptive statistics (means, standard deviations)
- Bonferroni corrected pairwise comparisons were used to examine differences in SS across the rare disease cohorts

Average levels of social support

Sickle Cell Disease (SCD)

- Sickle cell disease is caused by a change in genes that are involved in producing hemoglobin, an iron-rich compound found in RBCs.[4]
- While SCD may be invisible to the naked eye, healthcare providers can identify the condition using a number of tests, including newborn and prenatal screening, blood tests, and genetic tests.



Idiopathic Hypersomnia (IH)

- IH patients may fall asleep unintentionally or at inappropriate times, interfering with daily functioning. They may also have difficulty waking up from nighttime sleep or daytime naps.[6]
- Males and females are equally affected by idiopathic hypersomnia and the condition typically develops between 10-30 years of age.[7]
- The cause of idiopathic hypersomnia is not known. Some people with idiopathic hypersomnia have other family members with a sleep disorder such as idiopathic hypersomnia or narcolepsy.[7]

Alopecia Areata (AA)

- AA affects males and females equally and typically manifests during childhood.[5]
- The exact cause of alopecia areata is not known.
- An autoimmune mechanism is suspected in this disorder.[5]

Discussion

- Social support among women with AA and IH was lower than their male counterparts
- Compared to the other rare disease cohorts, women with IH reported significantly lower overall social support
- Women with IH fared worse in instrumental support when compared to those with SCD
- Emotional support was particularly low among women with IH when compared to those with AA

Conclusions

Why do women with IH receive less support?

Historically, women experience greater bias in healthcare, and subsequent underdiagnosis for many conditions. Clinical challenges to identifying and diagnosing "invisible" conditions, like IH, may be compounded by these social contexts, and contribute to feeling less supported.

Results

Description of patient cohorts

- Patients sociodemographic and health characteristics are described in **Table 1**
- The mean (SD) age of patients with SCD, AA, and IH was: 38.5 (16.5), 49.6 (16.6) and 40.7 (13.6) years
- More than half (>60%) of patients in each rare disease cohort were female
- SCD was most prevalent among patients who were African American (47.9%), whereas AA and IH were most prevalent among patients who were White (51.0% and 73.3%, respectively)
- Mean (SD) Charlson Comorbidity Index score[10] was 1.5 (2.6), 1.5 (2.7), 1.8 (2.9) for patients with SCD, AA, and IH, respectively

Comparisons of social support by rare disease

- Social support (any type) did not differ across male patients with SCD, AA, and IH, and average levels were higher than the sample average (i.e., all social support scores were >50).
- Female patients with IH had significantly lower social support scores when compared to other rare disease cohorts
  - Overall mean [SD] Social Support: IH was lower than both SCD and AA (53.0 [27.0] vs. 66.0 [28.4] vs. 63.3 [27.6], ps≤0.04
  - Mean [SD] Instrumental Support: IH was lower than SCD (53.7 [29.8] vs. 68.1 [29.6], p=0.04)
  - Mean [SD] Emotional Support: IH was lower than AA (52.3 [28.6] vs. 64.9 [28.3], p<0.01)

Table 1. Sociodemographic and health characteristics among patients with Sickle Cell Disease, Alopecia Areata, and Idiopathic Hypersomnia from the National Health and Wellness Survey

Sociodemographic and Health Characteristics		Sickle Cell Disease (N = 71)	Alopecia Areata (N = 600)	Idiopathic Hypersomnia (N = 105)
Age (in years), mean (SD)		38.5 (16.5)	49.6 (16.6)	40.7 (13.6)
Sex (Female)		46 (64.8%)	390 (65.0%)	65 (61.9%)
Race/Ethnicity	African American	34 (47.9%)	134 (22.3%)	7 (6.7%)
	Hispanic	20 (28.2%)	115 (19.2%)	13 (12.4%)
	Other	9 (12.7%)	45 (7.5%)	8 (7.6%)
	White	8 (11.3%)	306 (51.0%)	77 (73.3%)
Marital Status	Divorced, Separated, or Widowed	12 (16.9%)	124 (20.7%)	14 (13.3%)
	Married or Living with Partner	29 (40.8%)	344 (57.3%)	58 (55.2%)
	Single, Never Married	30 (42.3%)	132 (22.0%)	33 (31.4%)
Educational Attainment	4-year College Degree	15 (21.1%)	176 (29.3%)	27 (25.7%)
	Graduate Degree	10 (14.1%)	150 (25.0%)	26 (24.8%)
	High School or Less	23 (32.4%)	94 (15.7%)	20 (19.0%)
	Some College	23 (32.4%)	180 (30.0%)	32 (30.5%)
Employment Status (Employed)		41 (57.7%)	371 (61.8%)	63 (60.0%)
Health Insurance Status (Not Insured)		13 (18.3%)	75 (12.5%)	10 (9.5%)
Smoking Status	Current	23 (32.4%)	177 (29.5%)	39 (37.1%)
	Former	7 (9.9%)	129 (21.5%)	17 (16.2%)
	Never	41 (57.7%)	294 (49.0%)	49 (46.7%)
Alcohol Use (Drinks Alcohol)		49 (69.0%)	439 (73.2%)	72 (68.6%)
Exercised in the Past 30 Days (Yes)		48 (67.6%)	412 (68.7%)	65 (61.9%)
Charlson Comorbidity Index Score, mean (SD)		1.5 (2.6)	1.5 (2.7)	1.8 (2.9)

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