The Mental Health Burden of Rare Diseases Based on Insights From Patients & Caregivers Through RarePulse® Social Listening

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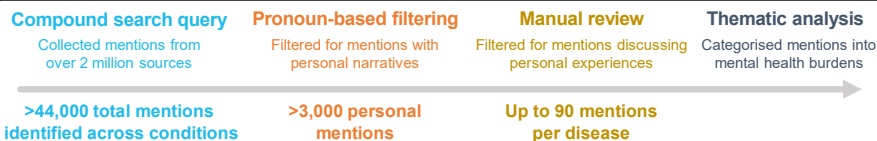
Objective

The mental health needs of individuals and families living with rare diseases are often under-recognised and under-served

This study aimed to systematically explore and compare the psychological & emotional challenges faced by individuals living with rare diseases and their caregivers, focusing on five diseases, **PKU, MPS, MSA, SMA, and CS**. RarePulse®, a large-scale social listening tool, was used to capture real-world experiences and unmet mental health needs

Methods

A structured, multi-stage social listening approach was conducted using RarePulse®, which leverages Brandwatch for data collection and sentiment analysis. The search strategy employed compound queries combining disease terms with mental health descriptors (e.g., "SMA" AND "Anxious") to capture mentions posted across multiple online platforms between 01/02/2024 and 01/02/2025. Mentions were manually categorised into themes to identify mental health patterns across the five rare diseases

Compound search query	Pronoun-based filtering	Manual review	Thematic analysis
Collected mentions from over 2 million sources	Filtered for mentions with personal narratives	Filtered for mentions discussing personal experiences	Categorised mentions into mental health burdens
>44,000 total mentions identified across conditions	**>3,000 personal mentions**	**Up to 90 mentions per disease**	

Results

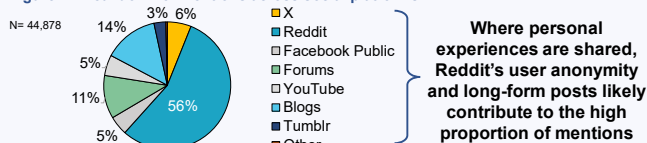### Highest mention volume across diseases was observed for CS

Figure 1. Volume of mentions across all diseases
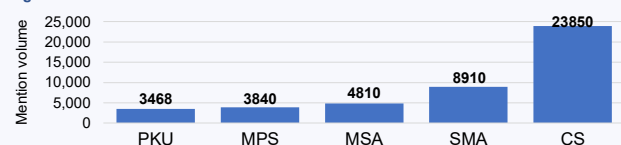
CS accounted for the highest number of mentions, likely reflective of a large and more engaged online community relative to the other diseases analysed. This could be related to diagnostic uncertainties and challenges associated with ongoing treatment monitoring associated with CS
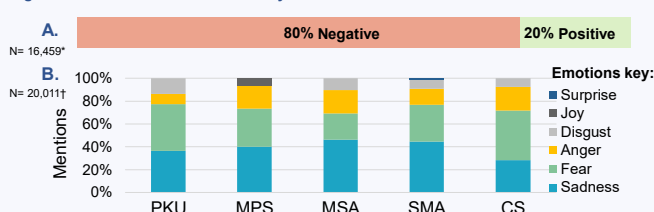
Mention volume across the five diseases predominantly originated from Reddit

Figure 2. Breakdown of mentions across social platforms

N= 44,878

Legend: X, Reddit, Facebook Public, Forums, YouTube, Blogs, Tumblr, Other

Where personal experiences are shared, Reddit's user anonymity and long-form posts likely contribute to the high proportion of mentions

Mentions were largely negative, driven by emotions of sadness and fear across all diseases

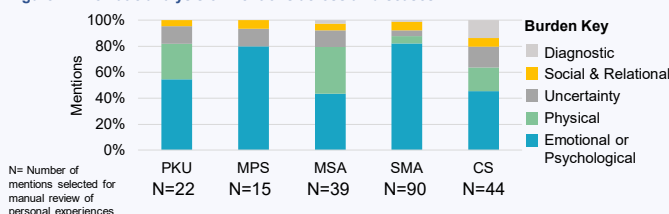Figure 3. Sentiment and emotion analysis of mentions across all diseases

A. N= 16,459*   80% Negative | 20% Positive

B. N= 20,011†

Emotions key: Surprise, Joy, Disgust, Anger, Fear, Sadness

Sentiment & emotion were determined via RarePulse®, leveraging Brandwatch

A. Sentiment analysis was possible on 16,459 mentions in the data set of which, 80% were considered as having negative sentiments across the diseases assessed

B. Emotion analysis identified sadness and fear as the most frequent emotions across all disease assessed

Negative emotional and psychological themes dominated mention volume across diseases

Figure 4. Thematic analysis of mentions across all diseases

Burden Key: Diagnostic, Social & Relational, Uncertainty, Physical, Emotional or Psychological

N= Number of mentions selected for manual review of personal experiences

PKU N=22, MPS N=15, MSA N=39, SMA N=90, CS N=44

Emotional and psychological themes were the leading category across all five diseases, though their weight varied. In SMA and MPS, over 80% of mentions were emotional or psychological, pointing to a heavy mental health burden

Various sub-themes of emotional and psychological challenges were identified, supported by the lived experiences of patients and caregivers

Table 1. Categorisation of mental health burden mentions

Sub-themes	Anonymised quotes
Grief	"I lost my [partner]…She's in my thoughts daily…I was her caregiver and the closeness that developed made her passing all the more traumatic… I still tear up at times when I think of her." **– Caregiver, online forum**
Anxiety and fear	"I'm still deeply afraid of having children in the future. If my partner carries the recessive gene, our child would almost certainly have this disease too, and that thought scares me a lot." **– Patient, YouTube**
Caregiver Exhaustion	"My [parent] basically got [Disease X]. It's come to the point where me and my siblings are all agreeing visitation dates to help out. All just feels a bit hopeless really." **– Caregiver, Reddit**
Life-ending thoughts	"[Disease X] has taken everything away from me. I had friends, I used to be able to drive and used to work a regular 40-hour schedule. All of it is gone…I pray to God every night to kill me." **– Patient, Reddit**
Isolation & loneliness	"I've been different since the day I was born…I tried to pretend it didn't bother me…growing up feeling like you're not normal in a society that values "normalcy"…It's extremely isolating." **– Patient, Facebook**

Conclusion

The social listening findings reveal that emotional and psychological burdens are widespread across PKU, MPS, MSA, SMA, and CS, with lived-experience narratives consistently describing high levels of negativity and distress

People frequently expressed grief, anxiety, fear, caregiver exhaustion, life-ending thoughts, and isolation, reflecting deep emotional strain that persists alongside physical disease management. Taken together, the consistency of these themes across rare conditions suggests a systemic gap in current care models, where psychosocial needs are insufficiently identified and supported

Study limitations[1]

Reliance on publicly available posts, potential sentiment misclassification, self-selection bias, and limited generalisability

The high proportion of mental health related themes was expected, as the search strategy applied filters to capture mentions of emotional and psychological challenges

Calls to action

RarePulse® social listening was able to uncover widespread emotional and psychological burden themes across PKU, MPS, MSA, SMA, and CS

This study shows that mental health challenges are not a secondary concern, but the stark reality of living with a rare condition. The overwhelming feeling of sadness, fear, and isolation across communities highlights the urgent need to protect individuals by bringing mental health to the forefront of rare condition care