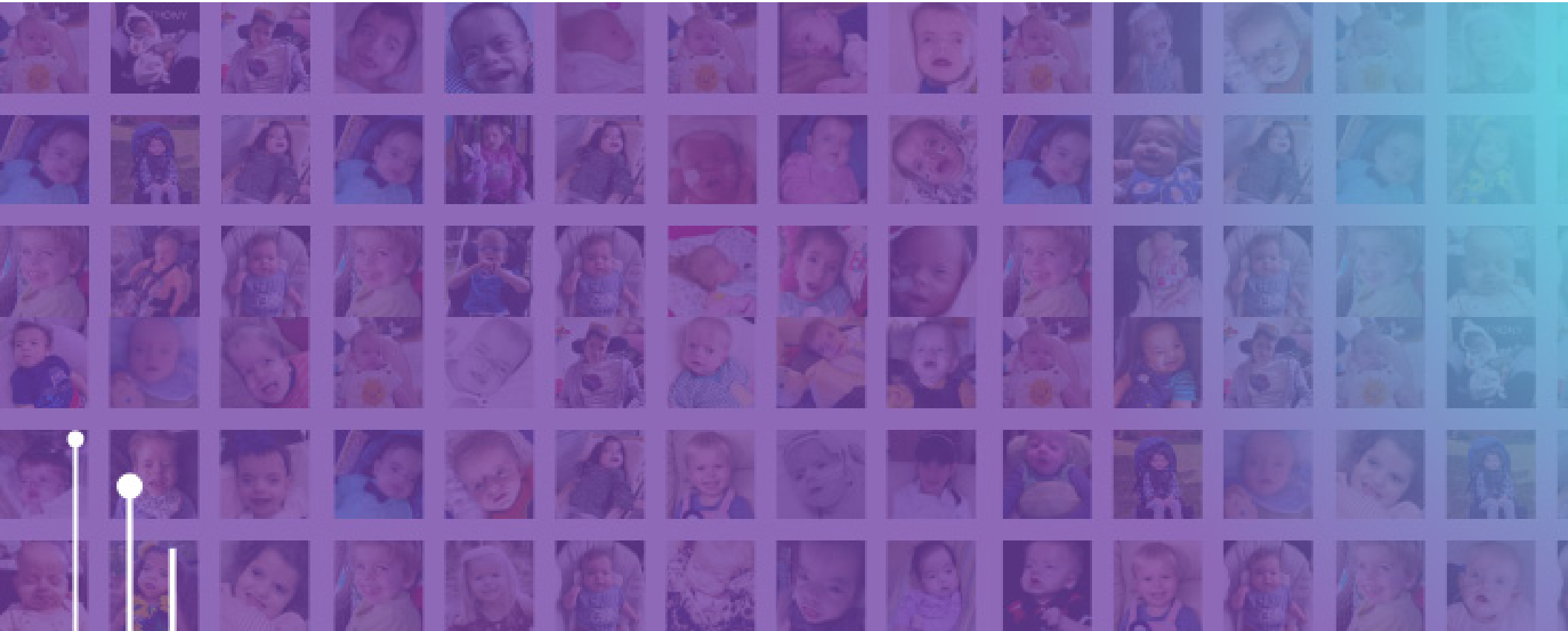


# Schinzel-Giedion Syndrome

## Community Voice Report

VOLUME 1, ISSUE 1 - SEPTEMBER 2024



DISEASE CATEGORY  
MULTISYSTEM



**TREND Community**<sup>™</sup>  
Turning Anecdotes Into Evidence<sup>™</sup>

# Actionable Community Insights

## The Role of Evidence in Progress

The pace of progress is always accelerating. Innovations in technology, combined with regulatory incentives, are helping biotech and pharmaceutical companies bring therapeutics and medical devices to patients faster than ever.

Unfortunately, most innovations are limited to finding treatments for diseases that are already well understood—that is, innovation in the rare disease space is lagging. In fact, 95% of the 10,000+ rare diseases have no approved treatments.

*True understanding of rare diseases* is the bottleneck. That's why the data and insights we capture from real patient voices are paramount to innovation. Our work unveils greater insights into disease presentation, symptoms, comorbidities, patient burden, and larger patterns. Ultimately, it is this holistic understanding that accelerates the development and approval of new therapeutics for which our communities are desperately waiting.

## TREND's Real-World Impact

From Community Voice Reports to peer-reviewed journals, the insights we glean from community conversations are translating to real-world impact. Visit our website to view posters, reports, abstracts, and other publications that we've co-created with our community partners.

*"The Community Voice Report helped us evolve from a Facebook group to a respected organization that is making a real difference in the HIE community."*

**—Betsy Pilon,**

*Executive Director, HOPE for HIE*

# How It Works

## Trusted, Invite-Only Partnerships

We establish trusted, long-term partnerships with invite-only online communities that are formed organically by people affected by rare, chronic, and emerging diseases.

## Proprietary Analytics

We harness machine learning and natural language processing techniques using Krystie™, our proprietary analytics engine that captures the perspectives and experiences of people within these trusted communities.

## Evidence Acceleration

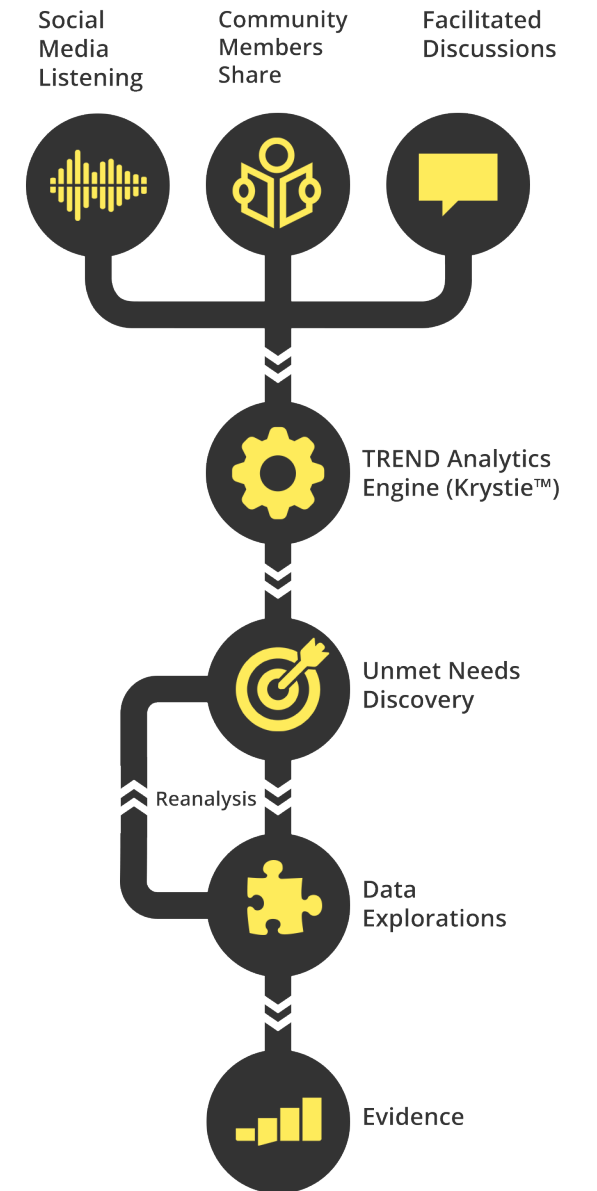
We discover insights on disease impacts and unmet needs. Our Community Voice Report and other real-world evidence can be leveraged for many different audiences and objectives.

## Sponsors Use TREND Data To:

- Capture insights earlier and faster
- Understand the patient/caregiver journey
- Launch relevant awareness campaigns
- Identify community leaders and influencers
- Discover underrecognized disease impacts
- Include the patient voice at all stages

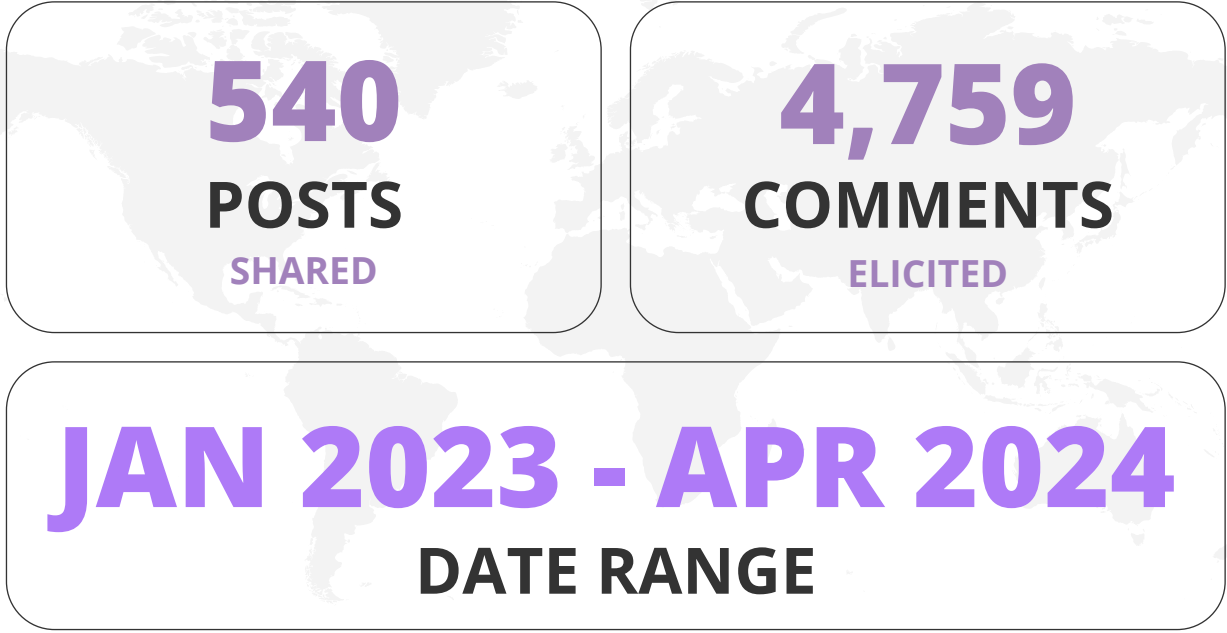
## Communities Use TREND Data To:

- Enable shared decision-making
- Strengthen advocacy efforts
- Inform FDA & other regulatory agencies
- Build communities' unique vocabulary
- Discover underrecognized disease impacts
- Ensure biotech and pharma hear their voices



# Analysis Background

Schinzel-Giedion Syndrome (SGS) stands as an exceptionally rare genetic disorder characterized by a complex array of physical, developmental, and neurological manifestations. Initially delineated in 1978 by Swiss pediatricians Albert Schinzel and Andres Giedion, the syndrome stems primarily from spontaneous mutations in the SETBP1 gene located on chromosome 18, with cases typically not inherited from parents. This gene's role in regulating gene expression and cell growth underscores its significance in the development and progression of SGS<sup>1</sup>. SGS is characterized by global neurodevelopmental impairment leading to moderate-to-profound intellectual disability, epilepsy (often refractory to treatment), hypotonia, spasticity, dysautonomia, hearing loss, and cerebral visual impairment. Other findings can include poor weight gain often associated with gastroesophageal reflux disease, chronic vomiting, constipation, gastroparesis, and/or feeding intolerance. Structural malformations can involve the heart, skeleton, kidney and urinary tract, genitalia, and brain. Anomalies of the liver, spleen, and/or pancreas are less common. Other features may include neuroepithelial neoplasia, severely disrupted sleep, choanal stenosis, inguinal hernia, sensitive skin, and increased risk of infection.<sup>2</sup>

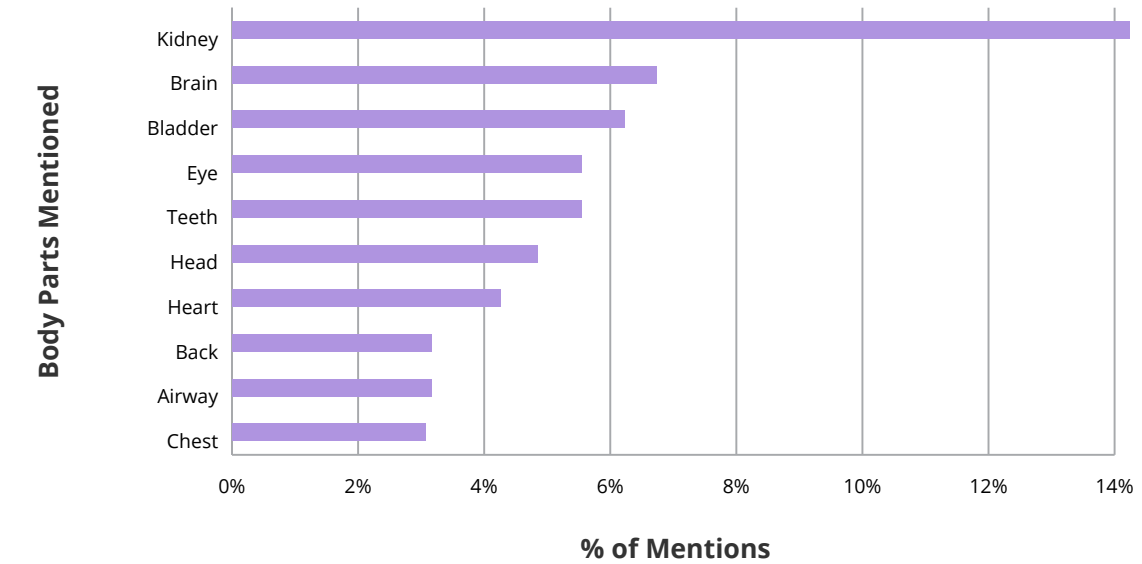
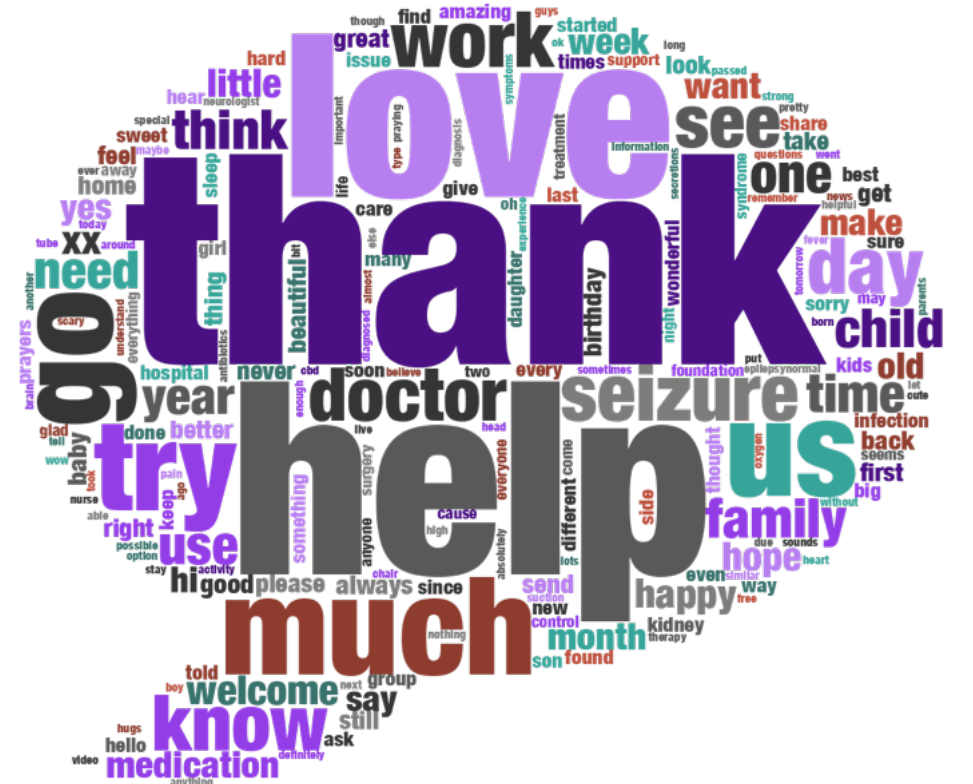


Although there exists no cure for SGS, management strategies predominantly center on providing supportive care tailored to address the individual's specific symptoms and needs. Given the rarity and complexity of the condition, a comprehensive approach involving geneticists, pediatricians, neurologists, and developmental specialists is paramount in ensuring holistic care and support for affected individuals and their families.

The mission of the Schinzel-Giedion Syndrome Foundation is to provide support for families caring for a child with Schinzel-Giedion Syndrome (SGS), raise awareness of SGS, and facilitate and support medical research.

To illustrate the most frequent words used by community members, a word cloud was generated. Some of the most frequent words included thank, help, and love. Words associated with medical experiences included family and child. Various words also denoted timing, such as day, month, year, and time.

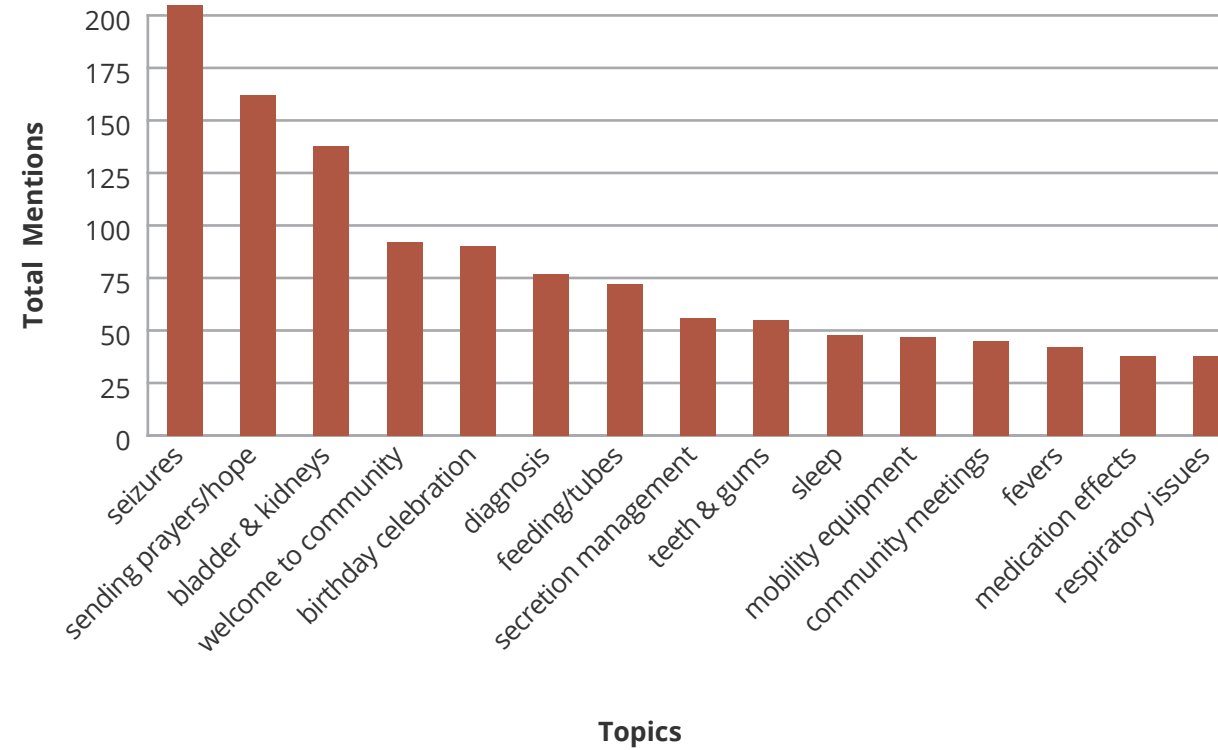
The most frequent body parts mentioned across posts and comments were extracted. The most frequent, Kidney, accounted for 14.6% of all body part mentions. Other internal organs included, Brain, Bladder, and Heart.



# Topics in the Community

Medical advancements have the potential to profoundly impact and reshape the characteristics of diseases over time. By actively listening to the community, we can gain valuable insights into these shifts when they happen. Our analysis involves clustering language that has similar meaning and occurs in similar contexts. This process is called topic modeling. The most frequent 15 topics are presented to the right. The largest topic was seizures (205 posts/comments), followed by sending prayers/hope (162 posts/comments). Topics in the middle included feeding tubes (72 posts/comments) and secretion management (56 posts/comments). The smallest topics, among the top 15 overall, included medication effects (38 posts/comments) and respiratory issues (38 posts/comments).

*It should be noted that the overall contents of the documents labeled "secretion management" (with top keywords "saline", "suction", "secretions", "mucus", and "chest") and "respiratory issues" (with top keywords "oxygen", "apnea", "sleep", "cpap", and "breathing") were distinct enough to warrant a separation of the topics.*

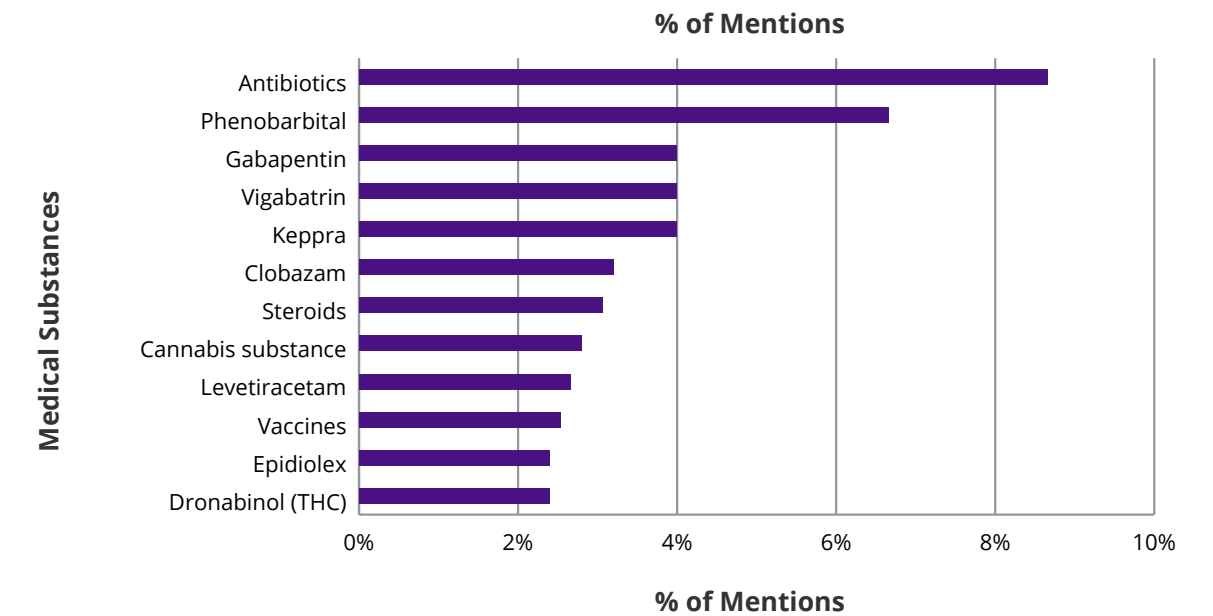
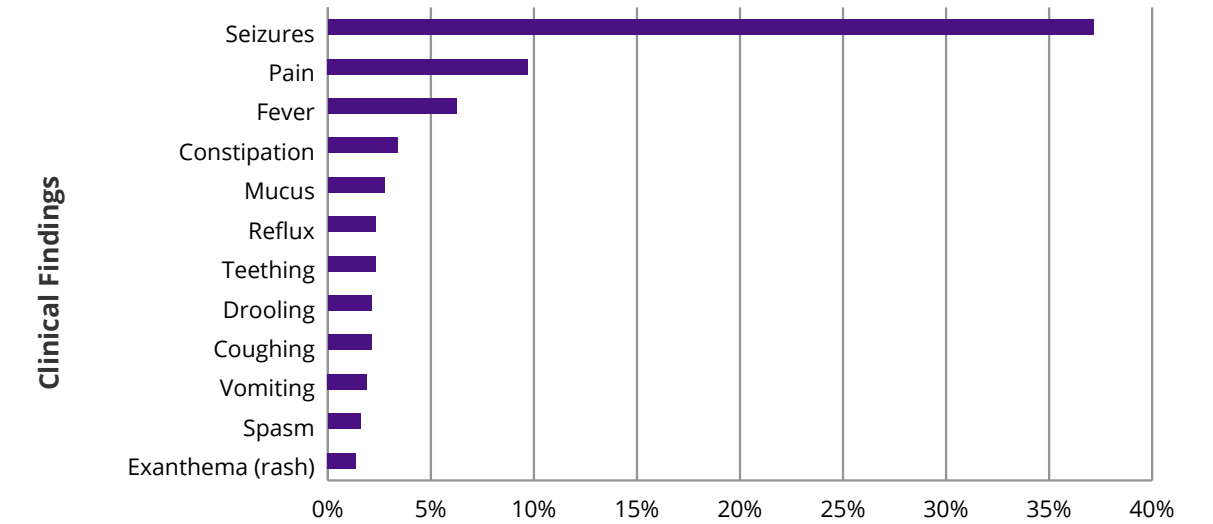


**“Always advocate. No one knows your baby better than you.”**

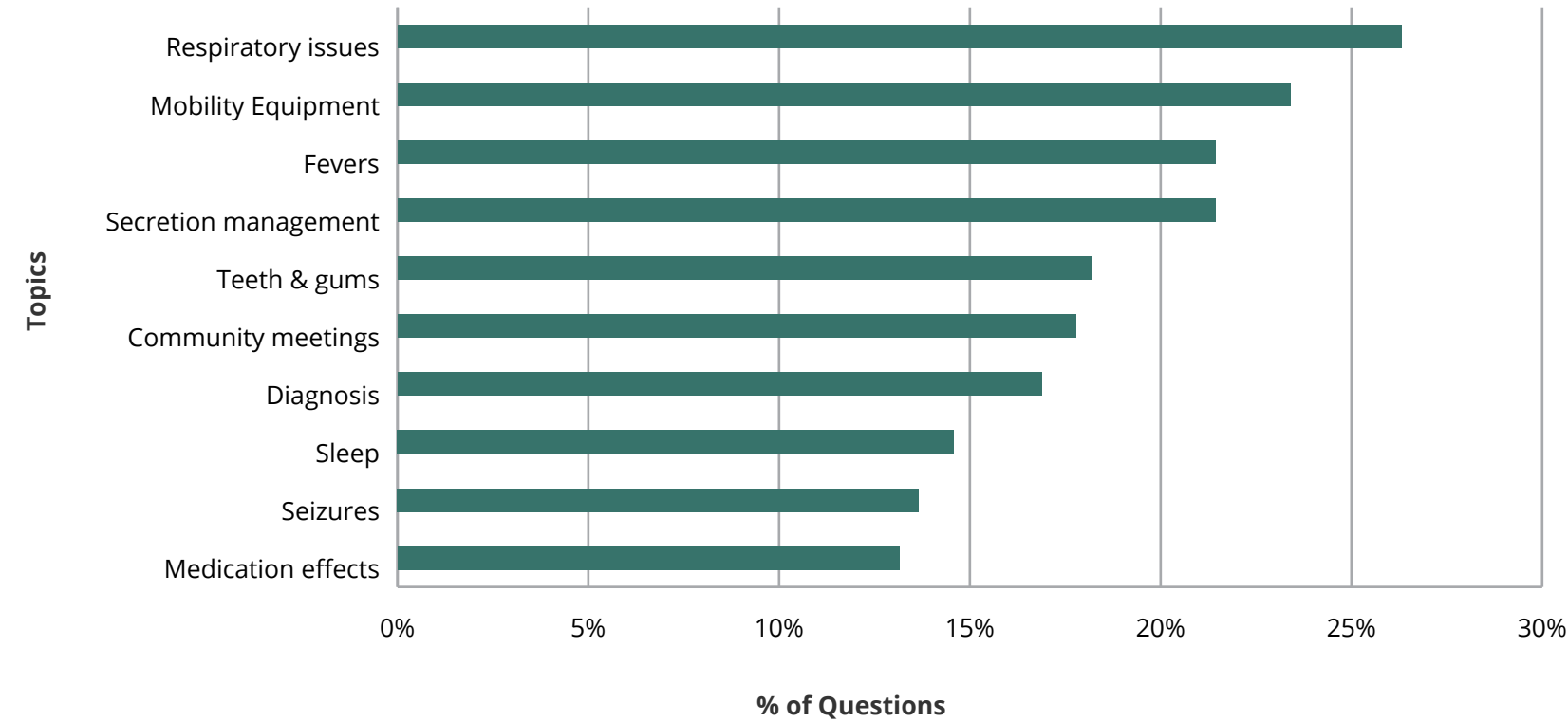
— Community Member

# Clinical Findings and Medical Substances

TREND's engine is trained to identify terms related to clinical language. Through this process, we can see the most frequently mentioned clinical concepts across clinical/medical domains. Two overarching clinical categories are presented here: Clinical Findings and Medical Substances. "Clinical Finding" refers to any sign, symptom, or other phenomenon related to a disease and its presentation. "Medical Substances" refers to any pharmaceutical medication and/or substance referenced for medicinal purposes. The top figure to the right illustrates the most common clinical findings (e.g., >35% of clinical findings mentions was seizures). The bottom figure to the right illustrates the most common substances discussed (e.g., >8% of medical substances mentions was antibiotics).

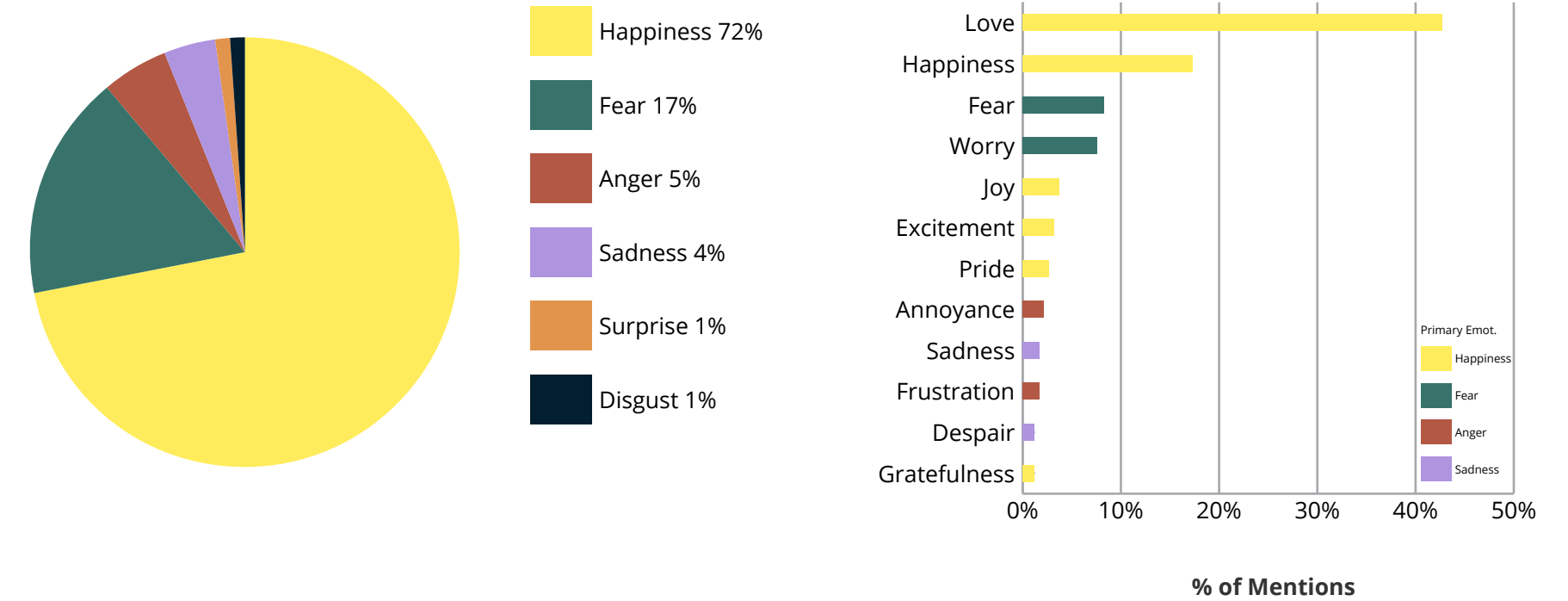


# Frequently Asked Questions



Community members often turn to their online support groups to find answers. Their questions can provide us with insights into the unmet needs of the community. For the most popular community topics (see Topics in the Community on page 6), we analyzed the proportion of questions included in conversations. This figure shows the topics with the largest proportion of questions. Specifically, the figure shows the highest frequency topics (y-axis) and the percentage of conversations that included questions (x-axis). For example, 26.3% of all conversations about respiratory issues involved a question. This list gives us insight into possible gaps in resources and the community's need for education and support.

# Emotion Classification

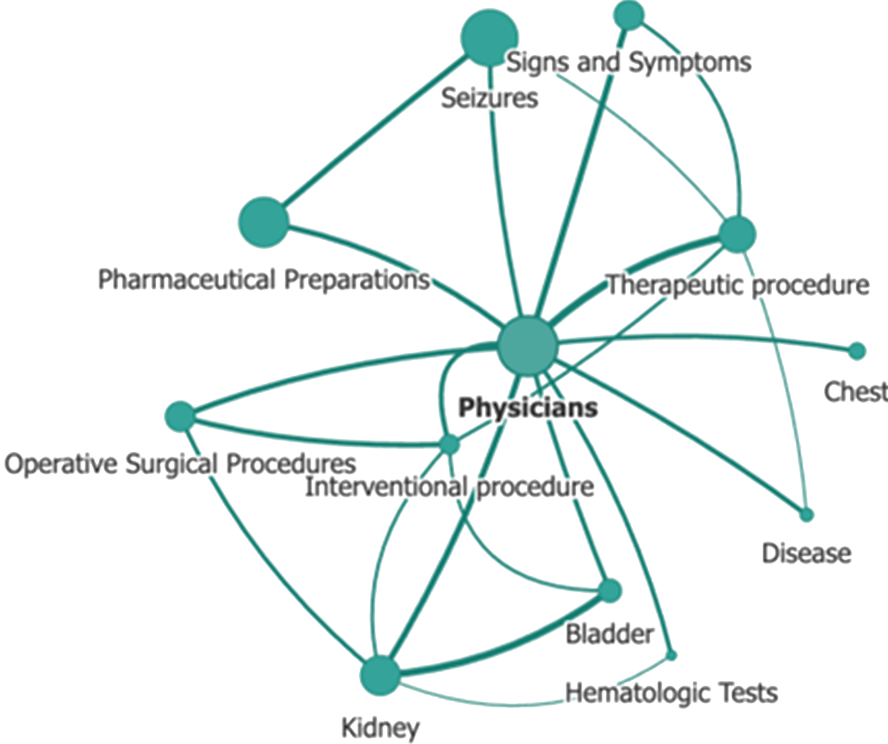


Krystie™ captures emotion language used by community members. After extracting emotions referenced, we can categorize them in six primary emotion categories (pie plot). We can then visualize the specific emotions mentioned in each category (bar plot). The most frequently expressed emotions fall under the categories of happiness and fear. Words and phrases associated with happiness included sending *love*, *speedy recovery*, *family*, and *hugs*. Words associated with fear included *concern*, *diagnosis*, and *hospital*.



# Physician Network Analysis

This figure reflects the Physician network in the SGS community. This network shows the concepts that co-occurred (i.e., occurred in the same post/comment) the most with the concept, Physicians (e.g., 'doctor', 'dr', 'physician', 'doc'). The larger the node (i.e., circle), the more the concept was mentioned. The thicker the edge (i.e., connecting line), the more the concepts were mentioned together. This provides a general indication of the clinical findings, testing, and management techniques that might be most frequently discussed with medical professionals. In this network, some prevalent categories included body parts (e.g., chest, kidney, bladder) and procedures (e.g., Operative Surgical, Therapeutic).



# Discovering Unmet Needs

### Seizures

Seizures were a consistent feature across various analyses.

### Management techniques

Both pharmaceutical and non-pharmaceutical management techniques were prevalent and had a high proportion of questions (e.g., mobility equipment, secretion management, medication effects). The community may benefit from specific resources regarding the best approaches for common day-to-day management.

### Navigating treatment

A diverse range of procedures was found in the Physician network. With so many options and potential interventions to consider, a substantial burden might be placed on caregivers as they navigate the range of management options available and coordinate care.

### Mental health

An encouraging finding in this report relates to the positivity in the SGS community. Many topics related to the concept of *community* (e.g., topics like *birthday celebrations, welcome to the community, community meetings*). Positive emotions were associated with community language (e.g., sending love/prayers). These findings suggest the SGS is a supportive, tight-knit group. Yet, it is important to note that more than 20% of emotion language related to *fear* and *anger* and topics included a range of themes relating to symptoms, treatments, and interventions. Rare disease burden and management creates a unique experience for caregivers that can negatively impact mental health. Increased support resources, outside the community, could benefit caregivers in their journey.

**“It feels like we are making major decisions every day.”**

— Community Member

**“I am so grateful to have found this group.”**

— Community Member

# Appendix

## Endnotes

- 1 "Home-Schinzel-Giedion Syndrome Foundation." *Schinzel-Giedion Syndrome Foundation*, 18 May 2024, [sgsfoundation.org/](https://sgsfoundation.org/).
- 2 Duis J, van Bon BWM. Schinzel-Giedion Syndrome. 2024 Mar 7. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. Bookshelf URL: <https://www.ncbi.nlm.nih.gov/books/NBK601394/>

# Acknowledgments

Thank you to the Schinzel-Giedion Syndrome community members for sharing their experiences. We are grateful to Nuala Summerfield and Dana Bradley for enabling access to the data used to create this report, providing their thought leadership, and amplifying the voices of people living with Schinzel-Giedion Syndrome by co-sponsoring this Community Voice Report.

## Sponsor



# About TREND Community

## Founders & Values

TREND Community™ was founded by the parents of a child with Prader-Willi syndrome who understand the needs of the rare and chronic disease community.

## We Lead With Empathy

We lead and listen with empathy toward each other, our partners, and our communities.

## We Amplify Voices

We believe that every voice has the power to make positive change.

## We Are Transformational

We believe that our approach has the potential to change the lives of people living with rare, chronic, and emerging diseases and alter the course of disease management for the better.

## Security

TREND secures all social data using state-of-the-art, private cloud servers. To protect community member privacy, conversation data from closed groups are anonymized, and the raw conversation data are never shared.

## Disclaimer

The researchers who prepared this report are not doctors, are not providing medical advice, and are only reporting what was said in the online conversations.

## IRB Exemption Status

Western Institutional Review Board determined that this study is exempt under 45 CFR § 46.104(d)(4).

## Quotes

All quotes were provided by consenting community participants through online surveys, one-on-one interviews, or online focus groups. Names are not included to protect participants' privacy.

## Data Ownership

Ownership of public posts or conversations from other social media platforms shared with TREND for analysis is subject to the social media platform's privacy policy, terms of service, and other applicable policies. TREND Community owns the results of our analysis and all other data and output we produce, including our Community Voice Reports.

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