THE OHIO STATE UNIVERSITY
COLLEGE OF DENTISTRY

Assessing Healthcare Experiences and Barriers to Care Among Individuals with Ectodermal Dysplasia

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BACKGROUND

- Rare diseases are uncommon individually, but collectively affect ~10% of the U.S. population. Individuals with rare diseases face many challenges:

Diagnostic uncertainty → Lack of knowledge → Limited providers → Financial constraints → Fragmented care

- **Ectodermal dysplasia (ED)** = heterogeneous group of hereditary conditions affecting development and/or homeostasis of two or more ectoderm derivatives, including hair, teeth, nails, and certain glands. ED has 49 known subtypes and no FDA-approved treatment.
- ED care is largely symptomatic, with dental needs comprising a significant burden among other specialist management.
- Extensive clinical/genetic research has been done, but insight on patient experience is largely based on anecdotes and case reports.

OBJECTIVES

Evaluate provider satisfaction and access to care

Compare medical versus dental care experiences

Identify areas for future research and/or support

METHODS

Population: Any English-speaking adult affected by ED and/or caregiver of a child with ED living in the North America.

Part A: Quantitative Data

- 76-item, ED-specific survey developed using validated instruments, rare disease literature, and expert opinion
- Recruitment via NFED database following pilot testing and review
- Descriptive analysis and comparison to reference data

Part B: Qualitative Focus Groups

- Series of one-hour, virtual focus groups using semi-structured interview guide specific to ED
- Recruitment from Part A, aiming to explain gaps and gain insight into the quantitative data
- Audio recordings transcribed and coded inductively using MaxQDA to develop codebook and identify key themes

RESULTS

Part A:

1.) Financial limitations on participants' ability to access care differed significantly for medical care versus dental care

2.) ED-affected individuals reported poorer oral health status and outcomes compared to national reference data

3.) All provider satisfaction measures (PSQ-18) differed significantly from the general population; financial domain also differed from rare disease population (Bogart et al)

4.) Self-reported adequacy of *medical* and *psychological* support matched other rare diseases; *dental, financial*, and *social* support domains differed

Part B:

"I love that I have doctors that have their eyes and ears open for new things, because my kid is not a one size fits all situation."

"Sometimes I feel like I have a part time job [...] like I'm phoning the ENT office, who then may phone the pediatrician here. And then three of her doctors are in the same building at the same hospital, but I have to send the same report separately to each of them."

"If you see or hear a condition you are not familiar with, listen to the patient more because you may not be familiar with it, but if we've been living with it, we can teach you."

Care Coordination
- Most responsibility assumed by individual/caregiver
- Value of academic centers and multidisciplinary teams

Oral Health
- Caregivers seeking action-oriented pediatric oral care
- High priority noting esthetics *and* function

Advocacy
- Role of self-advocacy in transitioning to adult care
- Eager involvement in research and policy efforts

Community
- Building resiliency through shared experiences
- Participants found benefit in focus groups themselves

Provider Interactions
- Positive: honesty, open-mindedness, listening, questioning
- Negative: rigidity, overconfidence, poor continuity

CONCLUSIONS

- Effective providers caring for patients with ED demonstrate empathy, curiosity, and willingness to collaborate with others on the care team
- Families affected by ED seek providers who demonstrate honesty about their knowledge, eagerness to listen, and an open-minded approach
- Financial constraints, particularly with dental care, pose a significant challenge for those with ED
- Fostering community support and skills of self-advocacy among ED-affected families is key

Improvement Areas Identified:

Provider Level

- Emphasize deliberative patient-provider relationships in medical and dental education
- Treat patients as a human, not a teaching opportunity, when involving research or education

Healthcare System Level

- Explore opportunities for formal care coordination
- Advocate for improved financial coverage (ELSA - Ensuring Lasting Smiles Act)

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