MAY 6, 2024

Summary Report

APBD & GSD IV
Focus Group

Strength & Power in
NUMBERS

APBD
ADULT POLYGLUCOSAN BODY DISEASE
RESEARCH FOUNDATION

HUMANIZED
SOLUTIONS

Index

Introduction

On May 6, 2024, the APBD Research Foundation hosted a virtual *Focus Group* that brought together, for the first time, over 50 participants representing the Adult Polyglucosan Body Disease (APBD) and Glycogen Storage Disease Type IV (GSD IV) communities. The participants included patients, family members, patient advocacy organization leaders, clinicians, and researchers from 13 countries.

The purpose of this meeting was to help our Foundation gather insights into the needs of the GSD IV community, collect best practices from both GSD IV and APBD families and representatives, and identify priority areas of possible near-term and future collaborations.

Partnering with Humanized Solutions, a Portugal-based company that supports healthcare stakeholders by implementing solutions co-created with the patient voice, the Foundation identified and connected with GSD IV families and community members living around the world. These individuals were then engaged in a series of discussions leading up to their participation in the *Focus Group*. A description of the methods employed can be found at the end.

Statement of Need

According to scientific literature, just 200 cases of APBD and 146 cases of GSD IV have been diagnosed worldwide. Given patient reports of their lengthy diagnostic odyssey and multiple misdiagnoses, we assert that both groups of patients are likely underdiagnosed.

A recently completed study by the Broad Institute of MIT and Harvard estimates the global genetic prevalence of all GBE1 diseases, including APBD and GSD IV, at 26,000. To advance therapeutic development, we believe it is imperative to identify, connect with, and build bridges between the APBD and GSD IV communities.

During the *Focus Group*, we heard from representatives of the APBD and GSD IV community about their diagnostic journeys and how it changed their lives. Parents of children with GSD IV and adult patients described their experiences, underscoring the heterogeneity of GSD IV symptoms, the spectrum of clinical involvement (hepatic, neurologic, muscular, and cardiac), severity, and age of onset. We also heard from researchers on a range of topics that highlight this meeting's narrative: the importance of building strength and power in numbers.

The *Focus Group* event serves as a launching pad for bringing two communities -- GSD IV and APBD -- together as we work towards therapies.

Key Findings and Recommendations

Patient/Family Needs:

- Connect with genetic testing and counseling services
- Connect with patient advocacy and research organizations offering educational and supportive resources
- Access to early recognition of symptoms as identified by the parent/care provider and referrals by primary care physicians to healthcare specialists (e.g., geneticist, neurologist, metabolic disease expert)
- Foster collaborations among healthcare specialists who are located internationally

Challenges in Diagnosis and Management:

- Misdiagnosis, underdiagnosis, and lack of awareness within the broader medical community
- Access to reliable information
- Specialist bias resulting in inappropriate management strategies
- Classification of genetic variants
- Understanding the natural history and progression of disease
- Conflicting prognoses provided by healthcare professionals to the family

The Role of Healthcare Professionals:

- Inform patients about patient advocacy organizations and support networks
- Adopt an international perspective and engage with global resources
- Maintain an open and investigative approach to facilitate diagnosis and care
- Collaborate with patient advocacy organizations to raise the public profile, provide continuing medical educational programs, and facilitate comprehensive support

Opportunities to Advance Therapeutic Development:

- Connect patients and families with existing research efforts around the world
- Collaborate on raising funds -- and offering scientific grant opportunities -- for research that advances both GSD IV and APBD
- Work together to identify funding opportunities for new programs
- Work to identify and address unmet needs
- Explore potential for international collaborations among researchers, patient advocacy organizations, and families
- Proactively seek out opportunities for collaboration among patient advocacy organizations
- Focus on making scientific information accessible
- Translate and simplify complex scientific information
- Finding, connecting with, and engaging new patients
    - Work with clinicians to distribute informational brochures from patient advocacy organizations
    - Leverage digital/social media platforms
    - Maintain a list serve of patients and families
    - Utilize communications platform to connect patients and families in real-time

Summary and Conclusion

This Summary Report compiles insights from the *APBD & GSD IV Focus Group* involving families, patients, and advocacy organizations. Key findings emphasize the importance of genetic testing, specialized healthcare professionals, support networks, natural history study and ongoing monitoring of disease progression, and collaborative efforts among specialists. Challenges include misinformation, specialist bias, and emotional distress from prognostic changes.

Effective disease management requires a comprehensive approach addressing medical, psychological, and logistical needs to improve patient and family well-being. Patient advocacy organizations play a crucial role in research engagement and international collaboration to enhance global research efforts. Enhanced international collaboration and proactive involvement of advocacy organizations are needed to fill gaps in educational resources.

In conclusion, we welcome the opportunity to collaborate with GSD IV and APBD community stakeholders. It is through strategic collaborations that we can gain momentum, build strength and power in numbers, accelerate research, and make treatments a reality.

Annexes

i) Methodology

The *Focus Group* encouraged participant interaction, yielding deeper insights and a nuanced understanding of the topics. The small group format allowed detailed exploration of opinions and behaviors, making it accessible to participants from all over the world. The moderated discussions enabled participants to share experiences and insights, suitable for the varied nature of the disease, promoting effective communication and wider participation.

Organizations, participants, moderation, and notes

- **Organizations:** Adult Polyglucosan Body Disease Research Foundation (APBD Research Foundation); Humanized Solutions
- **Participants:** patients, family members/caregivers, and representatives of patient organizations
- **Moderator:** Vanessa Ferreira, PhD, MBA
- **Notes:** Detailed notes captured participants' insights and contributions accurately

Three primary topics were explored:

1. **Experiences before, during, and after diagnosis**
    - **Genetic testing:** Crucial for accurate diagnosis and symptom management
    - **Specialized healthcare professionals:** Access to specialists significantly influences patient care
    - **Support networks:** These reduce isolation and provide practical support
    - **Continuous monitoring and early intervention:** Essential for effective symptom management
    - **Collaborative efforts among specialists:** Ensures comprehensive care

2. **Patient advocacy needs**
    - **Digital platforms:** Fundamental for initial contact and support

- **Conferences and summits:** Provide in-person interaction and access to healthcare specialists and researchers
- **Engagement and activity in support groups:** Varying engagement levels affect support quality
- **Personal connections:** Crucial for emotional and practical support

3. **Identifying potential solutions**
   - **Guidance towards patient advocacy organizations:** Healthcare professionals should guide families to patient advocacy organizations and support groups
   - **International perspective:** Engage with global experts to enhance care
   - **Open and investigative approach:** Vital for accurate diagnosis and management
   - **Collaboration with advocacy groups:** Essential for raising awareness and providing support

ii) List of questions used during the *Focus Group*

The session was structured to comprehensively explore three key thematic areas:

Part 1: Experiences Before, During, and After Diagnosis — This segment captured the participants' journeys from initial symptoms to diagnosis and subsequent life adjustments. The following questions were asked of family members/caregivers and patients:

- What interventions or support systems have been particularly effective in getting a diagnosis or managing your child's condition?
- Describe instances where you questioned the diagnosis or disease management process. How did these challenges impact your subsequent decisions regarding your child's healthcare?

Part 2: Patient Advocacy Needs — This segment focused on the support and resources needed by the community, evaluating the effectiveness of existing advocacy efforts and identifying gaps. The following questions were asked of family members/caregivers and patients:

- Where did you go to make your first connections with patient advocacy organizations and other families?
- How do you think healthcare professionals could help facilitate these connections?
- Can you describe some of the day-to-day challenges and 'secondary' symptoms associated with GSD IV that you feel are overlooked?

Part 3: Identifying Potential Solutions — Aimed at generating actionable solutions to improve patient care, advocacy, and community support, this segment facilitated collaborative problem-solving based on the discussions in the earlier segments. The following questions were asked of representatives from patient advocacy organizations:

- How do you find new patients/families impacted by a GSD IV diagnosis?
- Which digital tools are best for building community and sharing information about GSD IV?

iii) Resources

1) **APBD Research Foundation resources:**
- Monthly Newsletter Archive
- APBD Patient Registry
- NORD’s IAMRARE Registry
- Monthly APBD Patient Chats
- Monthly APBD Caregiver / Family Chats

2) **Other Glycogen Storage Disease Patient Advocacy Organizations:**

- ABGLICO Brazil
- AFG Association Francophone des Glycogénoses
- Asociación Española de Enfermos de Glucogenosis (AEEG)
- Associazione Italiana Glicogenosi
- Association for Glycogen Storage Disease - UK
- Association for Glycogen Storage Disease - US
- GlucoLatino
- Glykogenose Deutschland
- IsrAPBDf
- Metabolic Support UK
- Scandinavian Association for Glycogen Storage Disease

3) **Duke University Medical Center's GSD IV and APBD Natural History Study**

Acknowledgments

We extend our heartfelt gratitude to the participants of our *APBD and GSD IV Online Focus Group*. Your voices are vital in driving forward meaningful change and fostering a supportive community for everyone affected by these rare diseases. Thank you for your time, dedication, and openness.

A special thanks to the Humanized Solutions team for their support of this program. The company's co-founder, Vanessa Ferreira, PhD, MBA, was instrumental in guiding our efforts, connecting with, and engaging the GSD IV community, and preparing this Summary Report.

About the APBD Research Foundation

Founded in 2005, the APBD Research Foundation is the only US-based nonprofit supporting people with APBD and advancing research. As the trusted APBD hub for medical providers, the healthcare industry, and APBD families, we are the #1 source for patient information and support through our website, social media, e-newsletters, and patient and caregiver Chat events. Our mission is four-fold:

- Improve the diagnosis and treatment of APBD
- Support affected individuals and families
- Increase awareness of APBD among health professionals and the public
- Facilitate the translation of research into treatments and a cure for APBD

To learn more about APBD and our Foundation's work to find treatments and a cure, visit apbdrf.org.

APBD
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**Dedicated to finding a cure for APBD,
while improving the lives of those affected**

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