Pheo Para Alliance Patient-Centered Research on Challenges for those with Pheochromocytoma and Paraganglioma

Linda Rose-Krasnor[1], Stephanie Alband[2], Jacques W. M. Lenders[3], Lauren Fishbein[4]

[1]Pheo Para Alliance and Psychology Department, Brock University, St. Catharines, ON, Canada; [2]Pheo Para Alliance; [3]Department of Internal Medicine III, University Hospital Carl Gustav Carus, Technische Universität Dresden, Dresden, Germany and Department of Internal Medicine, Radboud University Medical Centre, Nijmegen, The Netherlands and Pheo Para Alliance Medical Advisory Board; [4]Department of Medicine, Division of Endocrinology, Metabolism and Diabetes, and Department of Biomedical Informatics, University of Colorado School of Medicine, Aurora, CO and Pheo Para Alliance Medical Advisory Board

About Pheo Para Alliance

Mission: to empower patients with pheo or para, their families and medical professionals through advocacy, education and a global community of support, while helping to advance research that accelerates treatments and cures.

Founded in 2007. Longest-standing and leading organization internationally recognized in advocacy for, and awareness of, pheo and para.

Scan of Pheo/Para Patient Experiences (SCOPPE) Survey

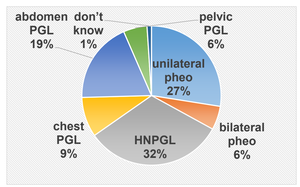
Goal: Explore the patient voice to verify, educate, explore connections and guide future path for the Pheo Para Alliance.

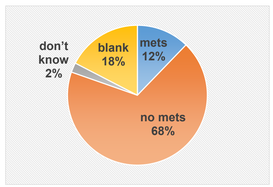
- Adapted from existing survey, based on consultations
- Clearance from a Research Ethics Board
- Recruitment through social media and email

Participant Characteristics

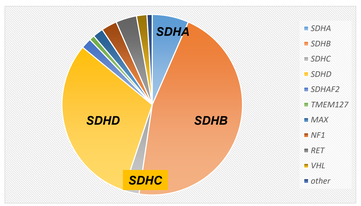
- 270 respondents
- Mostly female (81%), well-educated (76% with at least some undergraduate), white (88%), from urban/suburban areas (58%) in the USA (79%)
- Median age 52 yrs (SD 14 yrs)
- Most had PPGL (92%) and some were asymptomatic genetic carriers (8%)

Tumor Location and Presence of Metastatic Disease





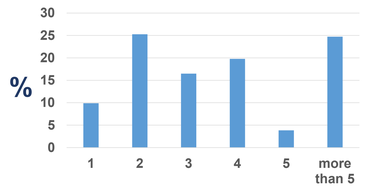
Clinical Genetic Testing



- Many participants had clinical genetic testing performed (78%)
- Of those, 60% had a known PPGL susceptibility gene pathogenic variant identified
- *SDHB* (48%) and *SDHD* (27%) were the most common

Delay in Diagnosis and Misdiagnosis are Common

- Median time from first symptom to diagnosis was 29 months (IQR 6-73.5)
- 48% saw ≥ 4 health care providers before being diagnosed



49% received one or more initial misdiagnoses

Anxiety	Depression	IBS
Diabetes	Hypochondria	Thyroid disorder
Menopause	Neuropathy	Other

Difficulties with Access to Care

- Many reported lack of access to an experienced medical team (29%) and to relevant information (26%) as well as poor communication among specialists (29%)
- 23% had to travel more than 100 miles and 36% more than 50 miles to be treated by a specialist
- Most (74%) would prefer to make decisions about their care in partnership with their medical team

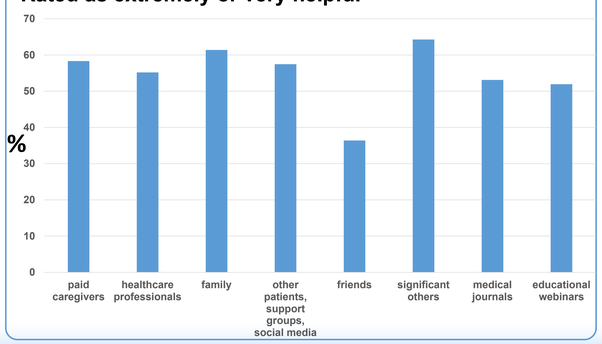
Important Aspects to Care

At least 75% of participants identified the following as "very" or "extremely important":

- More knowledgeable HCPs
- Better access to experts and medical centers
- More information about pheo/para
- Better medical team coordination

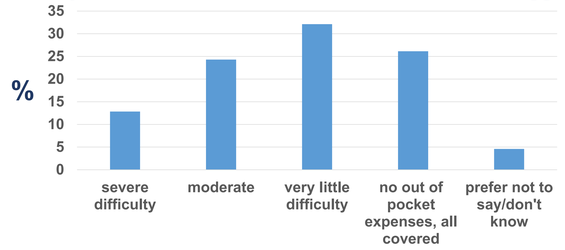
Many Sources of Support

Rated as extremely or very helpful



Financial Toxicity

In last year, >35% had moderate/severe financial struggle



Study Strengths and Weaknesses

Strengths

- Patient-initiated and designed survey
- Specific to PPGL
- Large sample size for this tumor type

Limitations

- Restricted sample
- Mostly white females in the USA
- Topics not covered (e.g., quality of life, coping mechanism)

Many Feel They are not Taken Seriously

"It's so rare, no one believes it, so I had to do all my own research and fight to get tests"

"Doctors not listening to patients. Assume we are wanting unnecessary tests….Assume the patient knows nothing or think we're just googling stuff."

"Drs just did not believe me – even with my blood & urine tests. Was told it was impossible for me to have it [pheo]"

Patient Struggles

"It's been very challenging, advocating for myself and pushing doctors and medical team to order the proper tests. There are symptoms I've experienced post surgery and post treatment that doctors don't have an explanation for."

"Since functional glomus jugulare tumors are rare, it is difficult to find recent studies that provide outcomes and longevity. The results of many studies are inconsistent and differ from one another. It is difficult to find someone to trust."

"Medical staff do not respect or acknowledge the knowledge of para pheo patients. We have to become experts in this field in order to navigate the lack of knowledge and care that the medical staff have."

"When I found out I had a glomus jugular tumor I didn't know who to talk to. No body had heard about this type of tumor, not even my doctors. I felt so alone."

"It sucks! Needs to be more awareness of the mental health impact this disease also has on both patient and family."

"Everything is always a fight with pheo/para no matter what country you are in. Tests take a long time to come back…. Doctors do not listen to us and dismiss half of our symptoms."

"My biggest concern is the fear of recurring paraganglioma and how to treat it if/when it happens."

"There is a real lack of answers on preventive protocol for my 3 children that I have passed the genetic mutation to."

"I work 4 jobs because I cannot afford to quit, can't work fulltime & do not qualify for social security benefits unless I quit working up to 1 year. Can't quit cuz I have bills to pay, there is no financial help from anyone."

"I am continuously saddened by the lack of clarity and info on head and neck Paras. It seems there's a focus on just hormonal tumors and evidence seems to suggest my head and neck tumors are relatively harmless. From my experience and my family's, this is untrue. I lost hearing, had a facial paralysis, have intracranial hypertension, nerve damage and damage from treatments. My cousin had her carotid torn and ended up in the icu. I wish there was a better focus on the risks associated with head and neck paragangliomas instead of the continued messaging that they are mainly asymptomatic especially on the pheopara site. My family and I feel very lost."

Patient Education and Advocacy Helps

"Thank you for continuing to raise awareness! My wife and I don't feel alone when we visit the alliance group page for information/guidance. Praying there will be more education done in this area to help me and others moving forward. Thanks for all you do!"

"I attended one of your webinars with a medical doctor/specialist presenting and found it extremely informative. Thank you!"

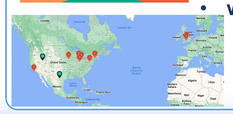
"The pheo para alliance has been a tremendous resource for me and my family. It has been a challenge to get answers and direction. So few experts."

Conclusions and Future Directions

- Delays, misdiagnoses and treatment inaccessibility are common and contribute to distress
- We can learn so much from our patients as we design surveys, clinical studies as well as for patient care.
- Sharing results with patients, HCPs, and researchers; incorporate findings into future research projects
- Help guide PPA strategic plan
- Repeat survey with more diverse sample and additional questions

Pheo Para Alliance Centers of Excellence

- Program meant to provide clear information for patients on where to obtain multi-disciplinary expert care
  - Clinical Center of Excellence
  - Clinical & Research Center of Excellence
- 12 centers so far (10 in USA; 2 in UK)
- www.pheopara.org/coe



Acknowledgements: Thank you to the participants in the survey