

Exploring the landscape of phenylketonuria education and learning needs of genetics trainees

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Introduction

- Phenylketonuria (PKU) is the most common inborn error of amino acid metabolism with an incidence in the United States (US) of 1 in 12,350 live births¹, making it one of the most commonly treated disorders in genetics practice
- Despite this, little is known regarding dedicated didactic and clinical PKU education provided by genetics training programs in the US
- In 2023, a convenience sample of six genetics trainees in the US were invited to participate in a 60–75 minute virtual interview and were compensated for their time
- Participants were asked to share their experiences and offer feedback regarding the PKU education they received during their post-graduate training
- Preliminary findings indicated the number of patients with PKU seen was variable (3–30 patients) depending on the program and year of training, and some trainees were involved in the medical management of PKU therapies
- The ability to follow patients longitudinally during training was noted to be challenging and there was mixed exposure to patients with PKU across the lifespan
- Interviewees reported receiving little to no specific didactic education on PKU with most of their education acquired via self-study
- Challenges were reported in keeping up to date on clinical practice guidelines, accessing updated resources, and reviewing recent literature for newly approved or emerging therapies
- A follow-up survey was conducted to explore the themes that emerged from the interviews with a new cohort of trainees and the findings are reported herein

Methods

- This IRB exempt survey was conducted in January 2024 to better understand current PKU education and clinical experience provided in US medical genetics training programs, gather information on PKU education needs and gaps in this population, and assess confidence of survey participants in managing patients with PKU
- Survey participants included current or recently graduated (≤ 1 year) medical genetics trainees in various programs accredited by the Accreditation Council for Graduate Medical Education (ACGME)
- A Microsoft Forms survey was developed and distributed to ACGME-accredited medical genetics program directors identified via ACGME database search for further distribution to intended survey participants

Results

- Demographics of the 21 survey respondents are presented in **Table 1**
- Respondents reported seeing a mix of patients with PKU from infancy to adulthood. On average, trainees had seen about 16 unique patients in clinic; however, they reported being directly involved in PKU management in less than half of this amount (**Figure 1**)
- The aspects of PKU care that respondents were most involved in included patient and caregiver education (86%, 18/21), newborn screening, adjustments to medical nutrition therapy, and adjustments to pharmacotherapy (81%, 17/21 each). There was less involvement reported with managing PKU symptoms and maternal PKU (57%, 12/21 each) (**Figure 2**)

- Most respondents received formal education on pathophysiology (90%, 19/21), medical nutrition therapy (86%, 18/21), burden of illness (81%, 17/21), and pharmacotherapy (76%, 16/21) as part of their training, while 2 participants (10%) reported no formal PKU education
 - Amongst the survey participants who received pharmacotherapy didactics (16/21, 76%), the majority were at institutions actively participating in PKU research (12/16, 75%)
- The majority of respondents (81%) agreed or strongly agreed that their training program's focus on PKU was comparable to that of other inborn errors of metabolism (IEMs)
- Overall, respondents were more comfortable with dietary management for PKU (81% were somewhat, fairly, or completely confident) compared to pharmacotherapeutic management (52–67%)
- Almost 40% of respondents were unfamiliar or somewhat unfamiliar with the American College of Medical Genetics & Genomics (ACMG) clinical practice guidelines for PKU (**Figure 3**)
- Most (18/21, 86%) respondents reported that they anticipate treating patients with PKU after training; of those, 78% (14/18) felt adequately prepared to directly manage these patients, while 22% (4/18) did not. Furthermore, most (81%, 17/21) reported they would personally benefit from receiving additional PKU education supplementary to their training (**Figure 4**)
- **Figure 5** summarizes the aspects of PKU education considered most critical to improving confidence in clinical management

Table 1. Demographics of survey respondents

Question	Summary of Responses (n=21)
Training Program*	Medical Genetics & Genomics (n=9) Pediatrics/Medical Genetics & Genomics (n=8) Medical Biochemical Genetics (n=3) Clinical Biochemical Genetics (n=1)
Status	Current Trainee (n=10) Recent Graduate (≤ 1 year) (n=11)
Institutional involvement in PKU Research	Yes (n=12) No (n=4) Not Sure (n=5)

*There was fair geographic distribution across the US for respondents who self-reported their institution.

Figure 1. Patients with PKU seen vs. directly managed during training

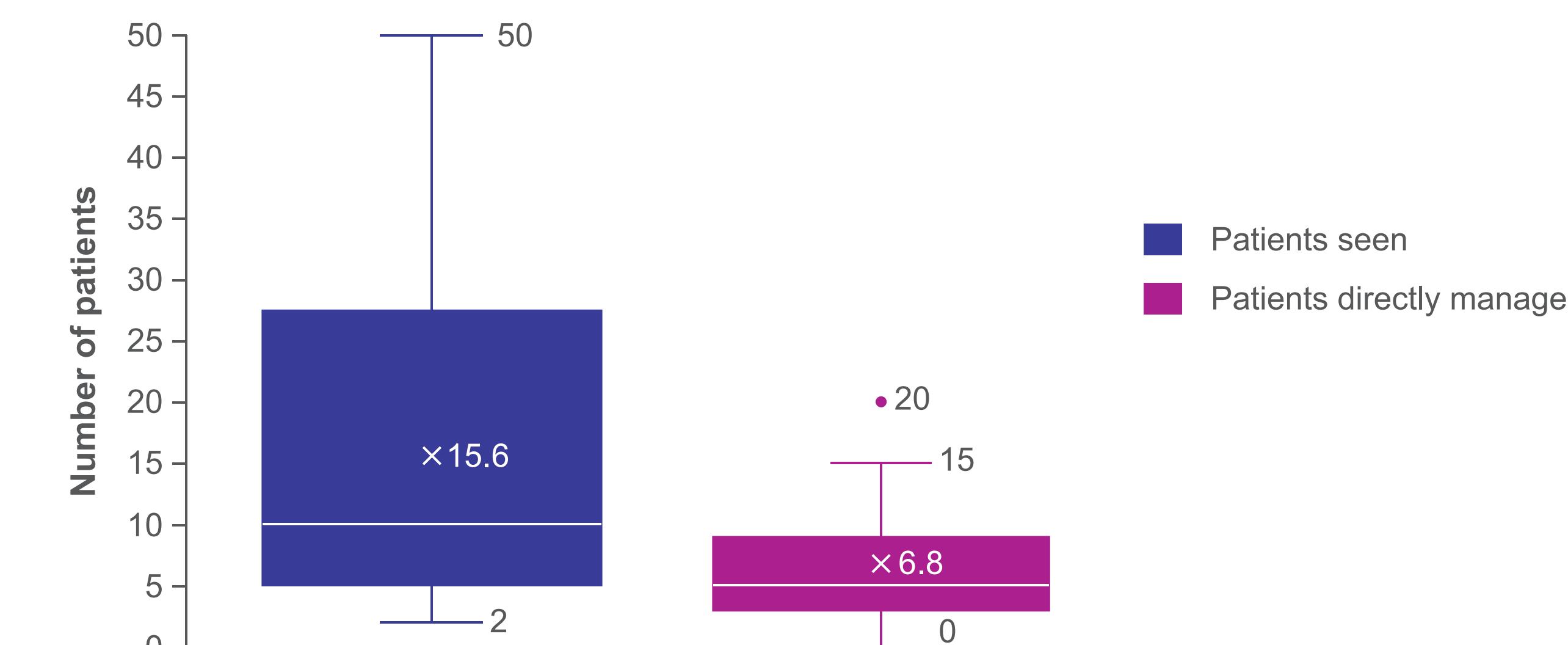


Figure 2. Involvement in aspects of PKU diagnosis and patient management

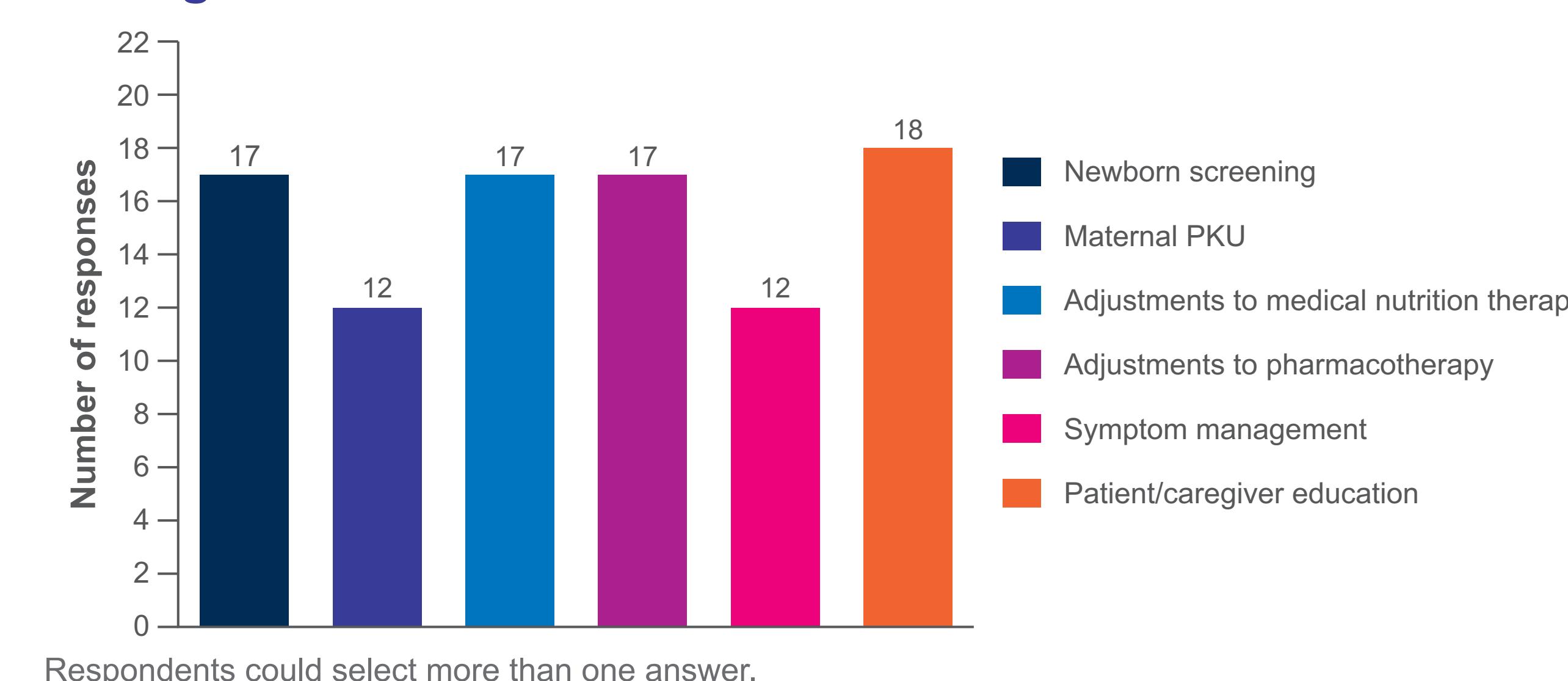


Figure 3. Level of familiarity with ACMG clinical practice guidelines for PKU

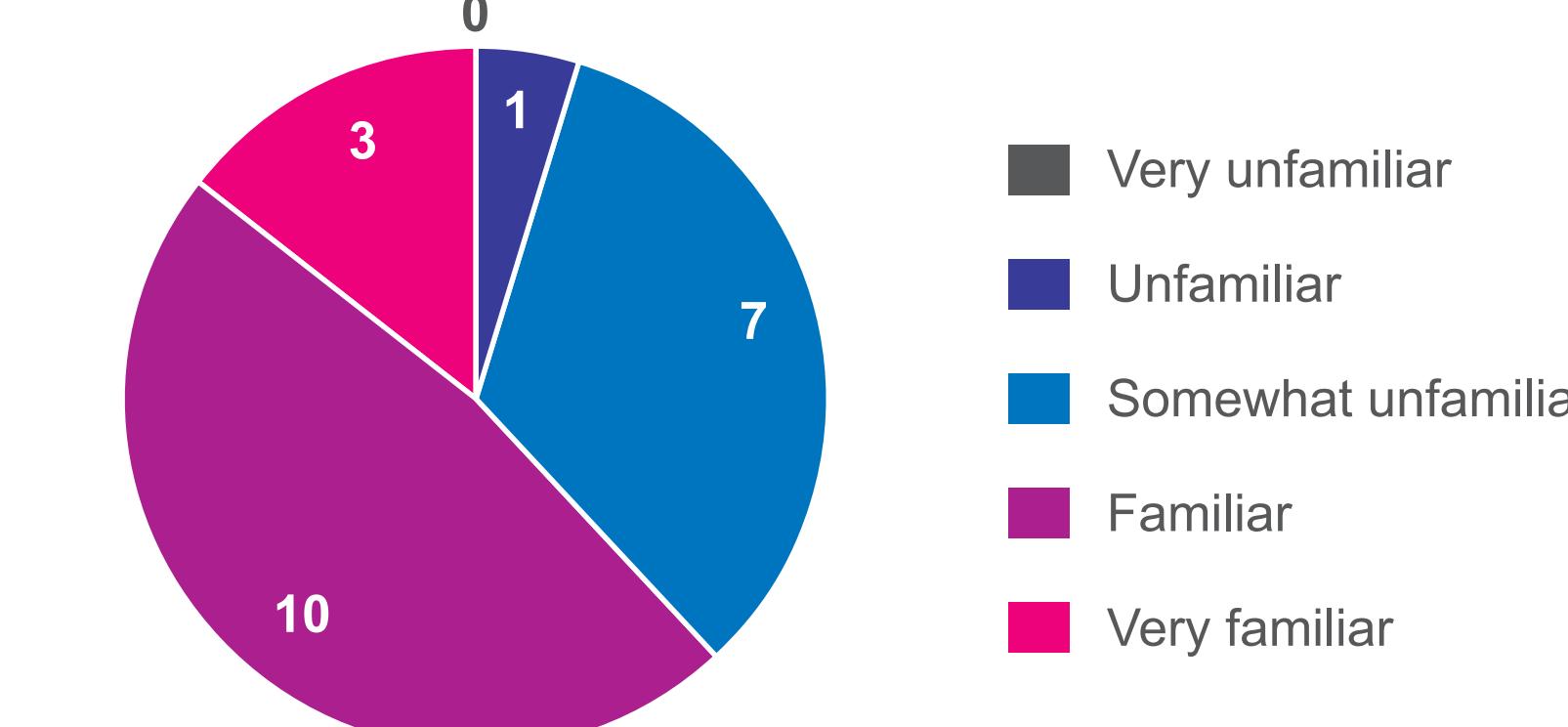


Figure 4. Participants who reported they would benefit from additional PKU education supplementary to training

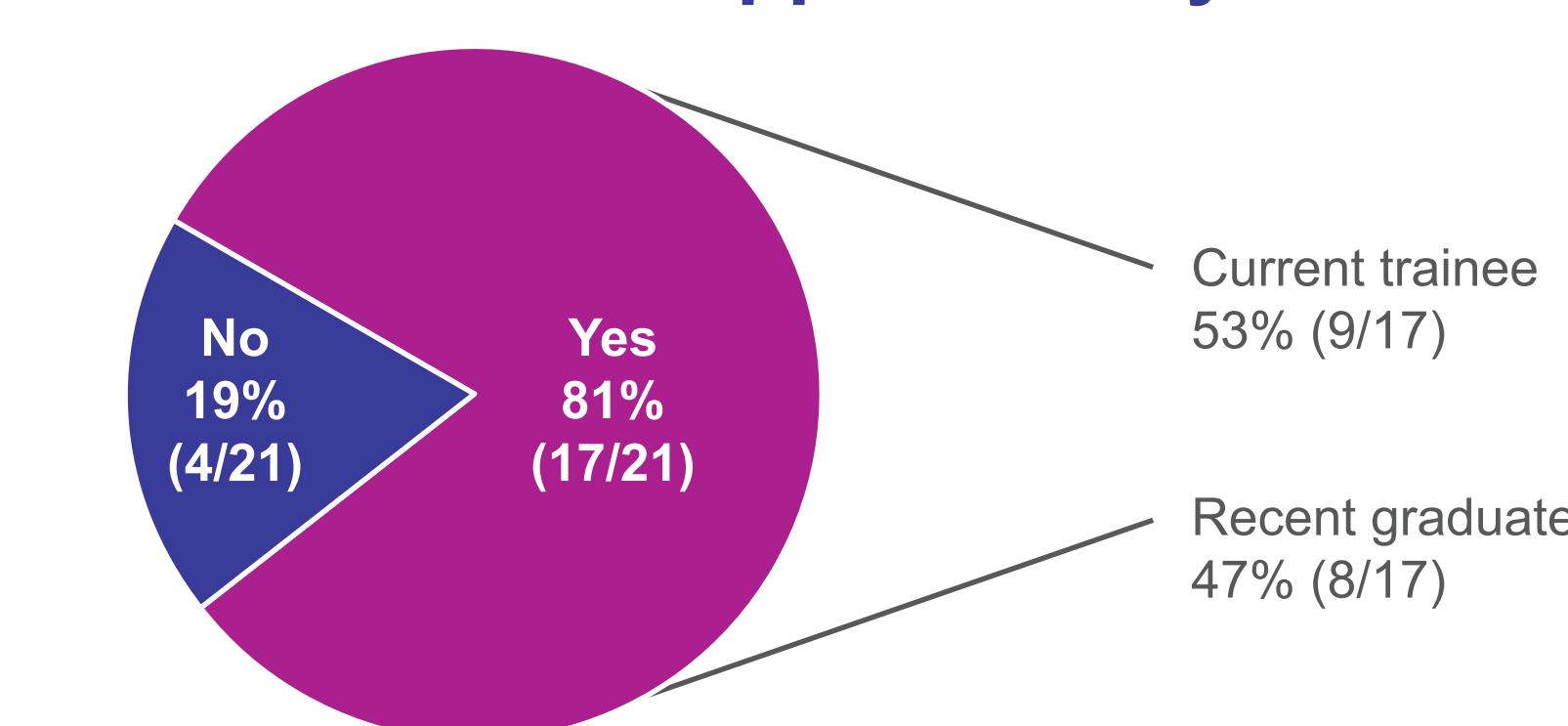
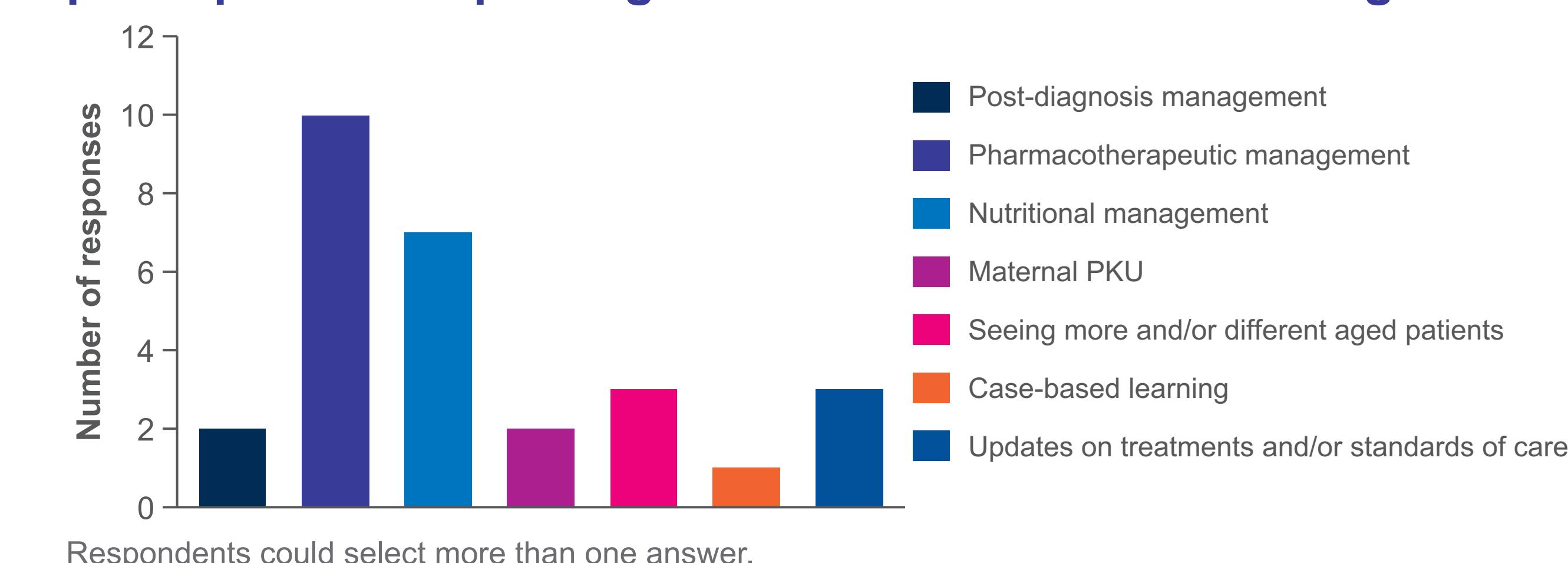


Figure 5. Aspects of PKU education considered most critical by participants in improving confidence with clinical management



Conclusions

- Qualitative interviews in a small group setting suggested the PKU education received by genetics trainees in the US differed in areas including nutritional and pharmacotherapeutic management, as well as patient exposure across the lifespan
- Survey responses from 21 trainees or recent graduates confirmed variable exposure to PKU patients seen and managed during training. While in general, respondents were confident with the clinical management of patients with PKU, several gaps in knowledge were reported (e.g. Maternal PKU, PKU symptom management)
- The mixed responses regarding familiarity with the ACMG clinical practice guidelines for PKU highlights an opportunity for increased exposure
- Regardless of comfort level with managing PKU, additional education was desired regarding nutritional and pharmacotherapeutic management to improve confidence
- There is a need to provide more post-graduate PKU education, given that it is the most common IEM seen in clinical practice; supplementary education may improve clinician knowledge and confidence
- These findings may extend to other IEMs given the focus during training was reportedly comparable

References

1. Foreman PK et al. *Orphanet J Rare Dis.* 2021; 16:253.

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