

No Turning Back: How to Support the Mental Health Needs of Families with PKU

Dr Nadirah Damseh

Dr Raed Al-Hindi



Outline



The Rebuilding Alliance PKU Tiger team

What is PKU?

- Background
- Screening
- Required treatment

PKU in Palestine

- Formula, newborn testing
- Obstacles
- Bigger picture

PKU Tiger Team

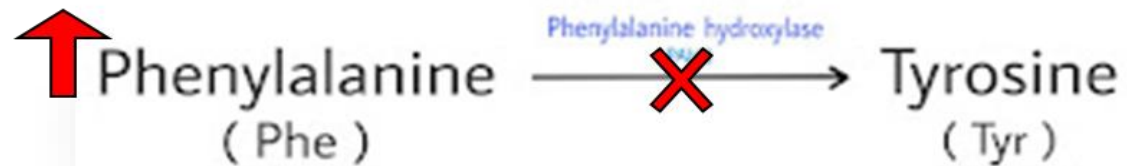


- 4 continents
- Medical professionals
- Volunteers
- Individuals with PKU



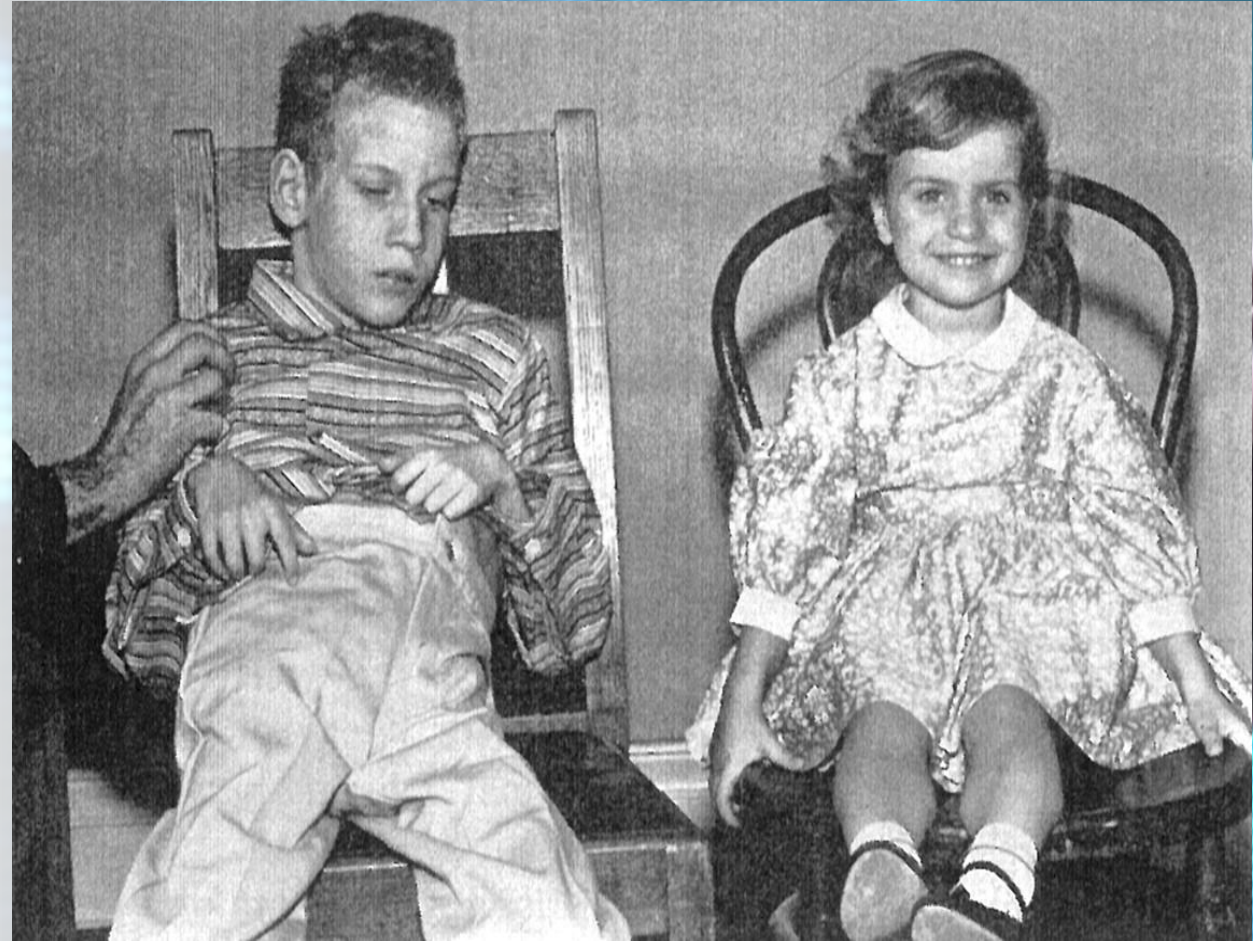
Phenylketonuria

- The most common preventable cause of intellectual disability.
- Results from deficiency of a liver enzyme called Phenylalanine hydroxylase.
- This enzyme deficiency results in intolerance to the dietary intake of an essential amino acid called phenylalanine.
- Accumulation of phenylalanine produces a spectrum of medical complications.



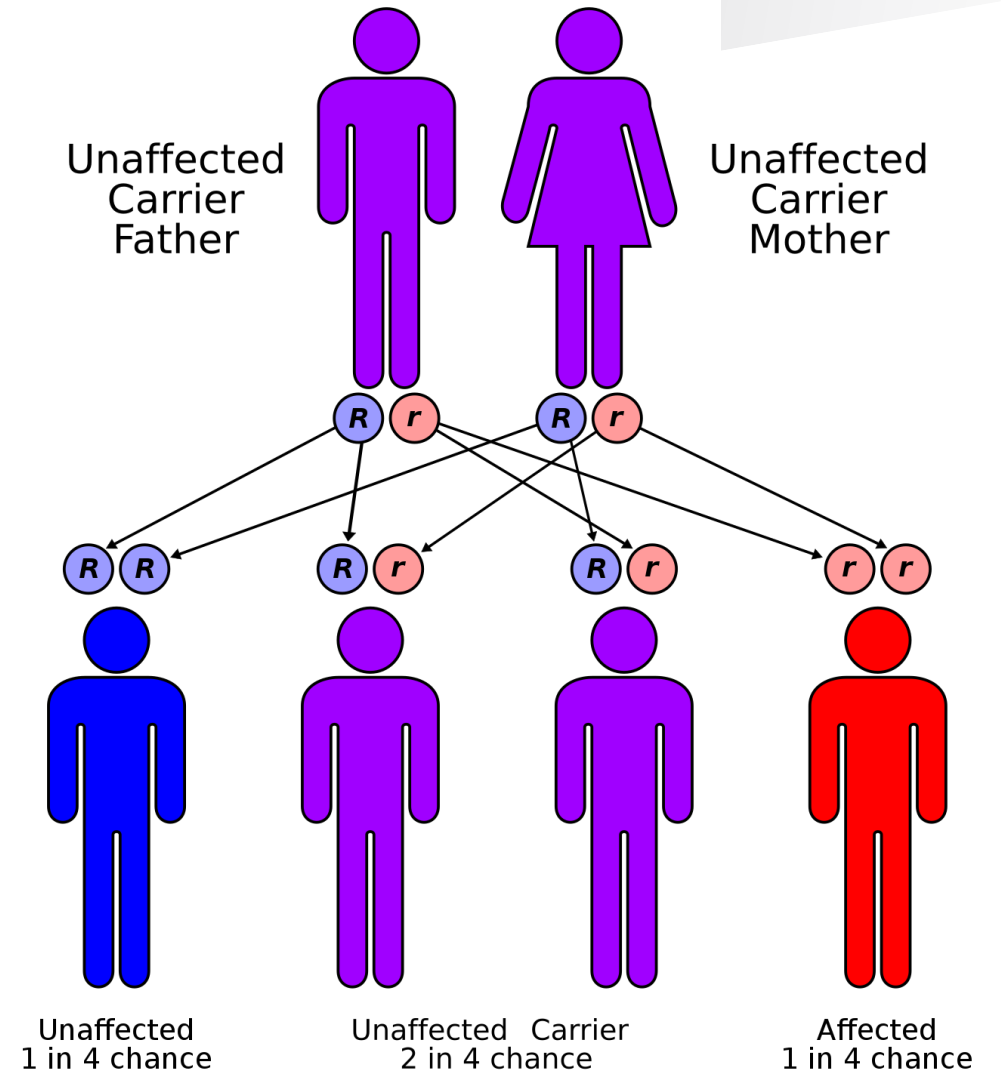
Clinical features if untreated

- Irreversible brain damage and intellectual disability
- Neurological issues: seizures and tremors
- Behavioral, emotional, social problems
- Early death



Genetic Basis of PKU

- Autosomal recessive pattern of inheritance
- Typically both parents are carriers
- Children / adults with PKU never “grow out” of the condition



'Golden Standard' of Care



1. Newborn screening
2. Medical formula / diet
3. Regular Phe level testing





Management

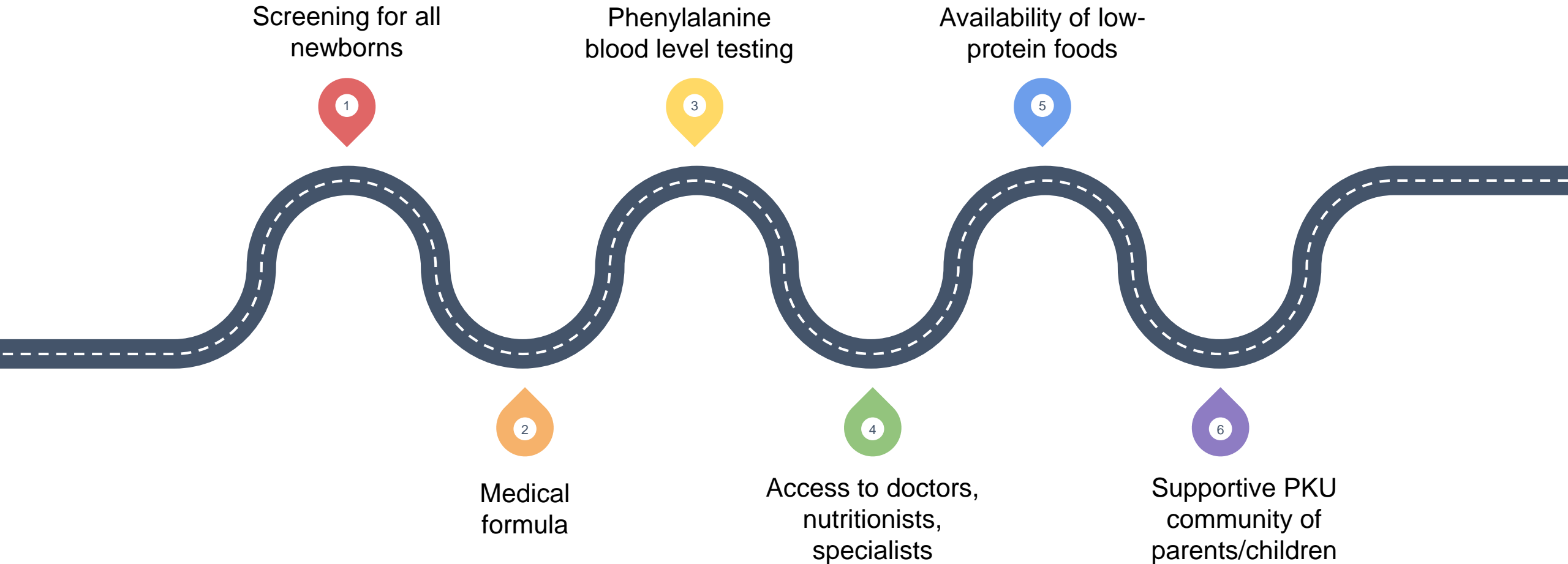
- PKU is a treatable condition.
- Restriction of dietary phenylalanine (PA) using low-protein food and Phenylalanine-free medical formula
- Treatment has to be initiated as soon as possible after birth to achieve PA levels of 120-360 $\mu\text{mol/L}$ (2-6 mg/dL).



Phenylketonuria in Palestine

- Background Population : 5.24 million. 61% West Bank, 39% Gaza
- Birth Rate : 397 per day
- Over 580 children in Gaza & the West Bank suffer from Phenylketonuria (PKU).

What does sustainable care require?



Challenges

Occupation
and border
restrictions

Government
priorities

Financial
burdens

Limited
resources

Lack of data

Limited
management
/treatment





Newborn screening in Gaza

- Screening Program in Palestine began in 1994.
- Inconsistent newborn screening over the last few years
- Only location for testing: Ministry of Health Central Laboratory
- Lab damaged in May bombing
- 60,000 births/year in Gaza alone

Shortage of PKU test materials threatens Gaza newborns

Failure to conduct test of phenylketonuria leaves the lives of Palestinian newborns at risk

Nour Abu Aisha | 26.01.2021





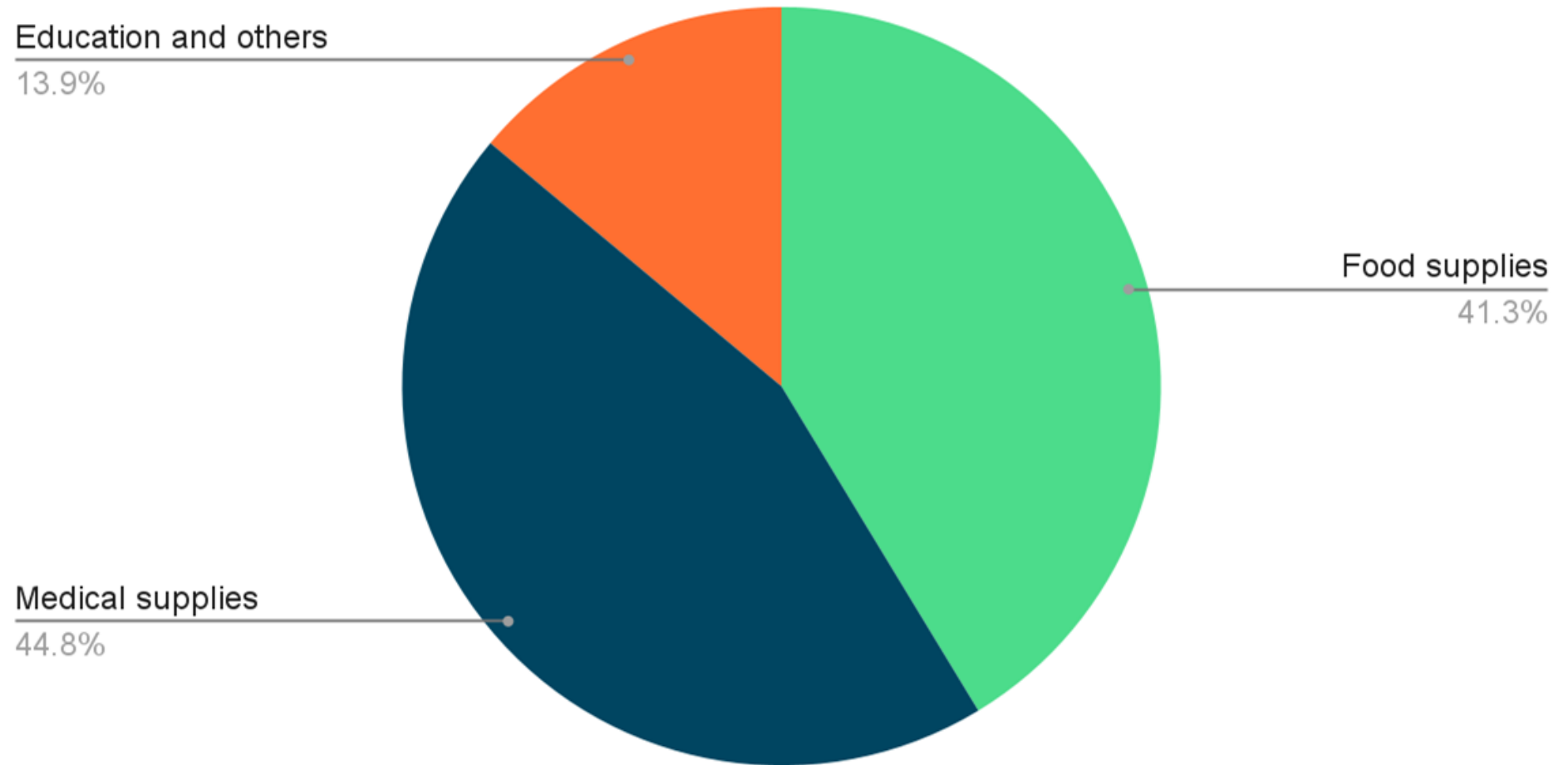
PKU children need regular
and secured supply of
specialized medical food

Gaza PKU Parent Survey



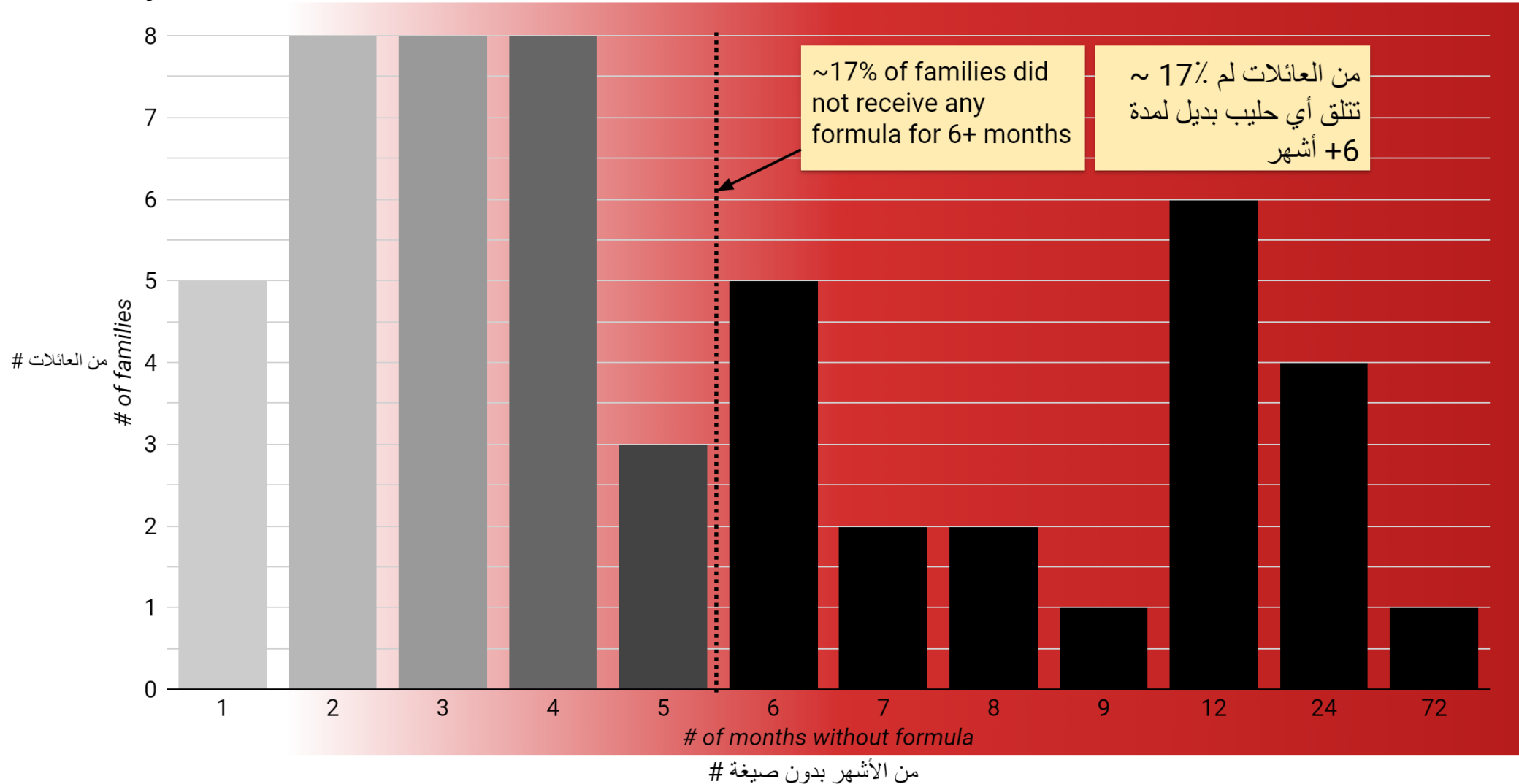
- Dec. 2020
- 126 respondents
- 36 questions
- Understand situation and community needs

Please let us know how we can help you and other families who have children with PKU.



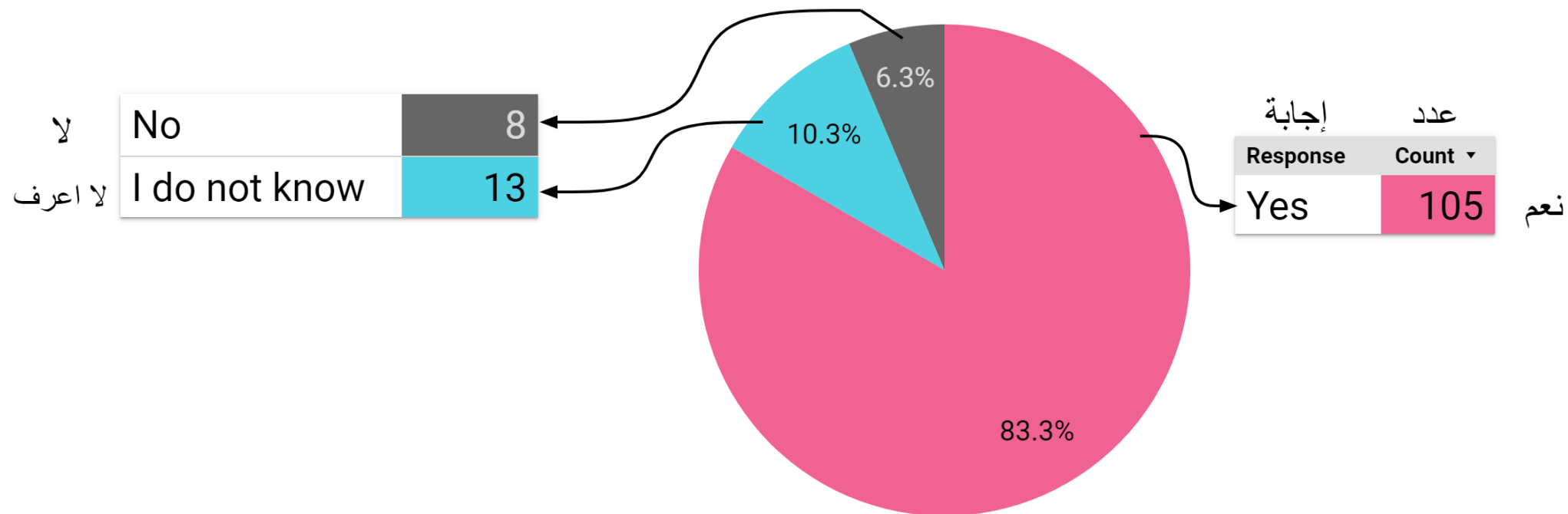
The chart below displays the number of months a family had gone without receiving any formula.

يعرض الرسم البياني أدناه عدد الأشهر التي مرت بها العائلة دون تلقي أي صيغة.



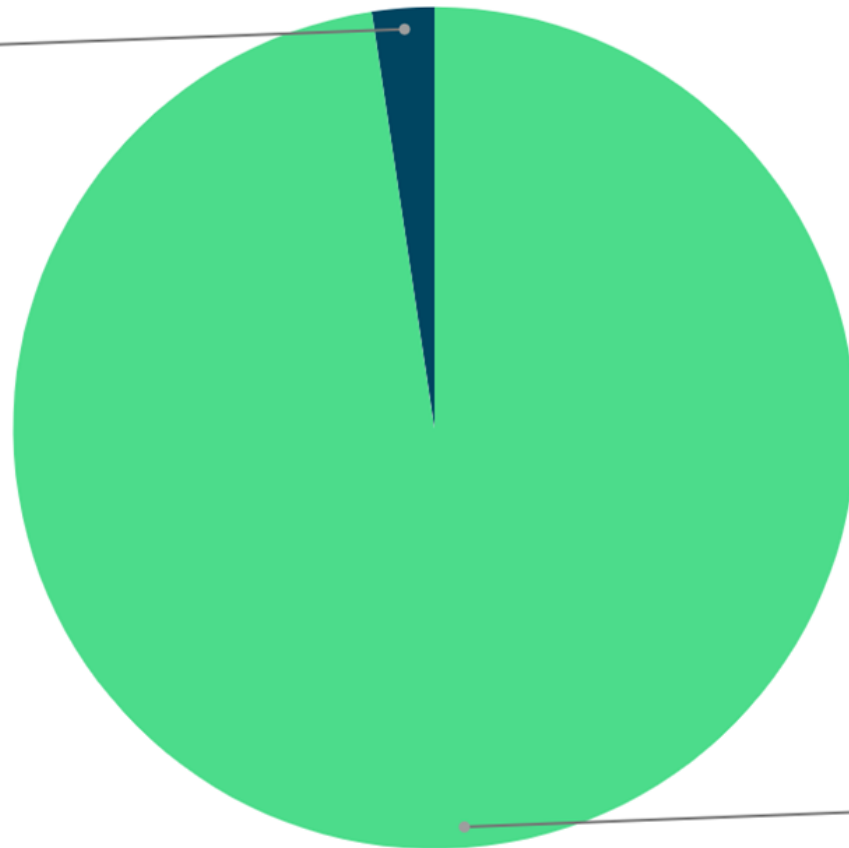
Is your child being tested for phenylalanine levels?

هل يتم اختبار مستويات الفينيل ألانين لطفلك؟



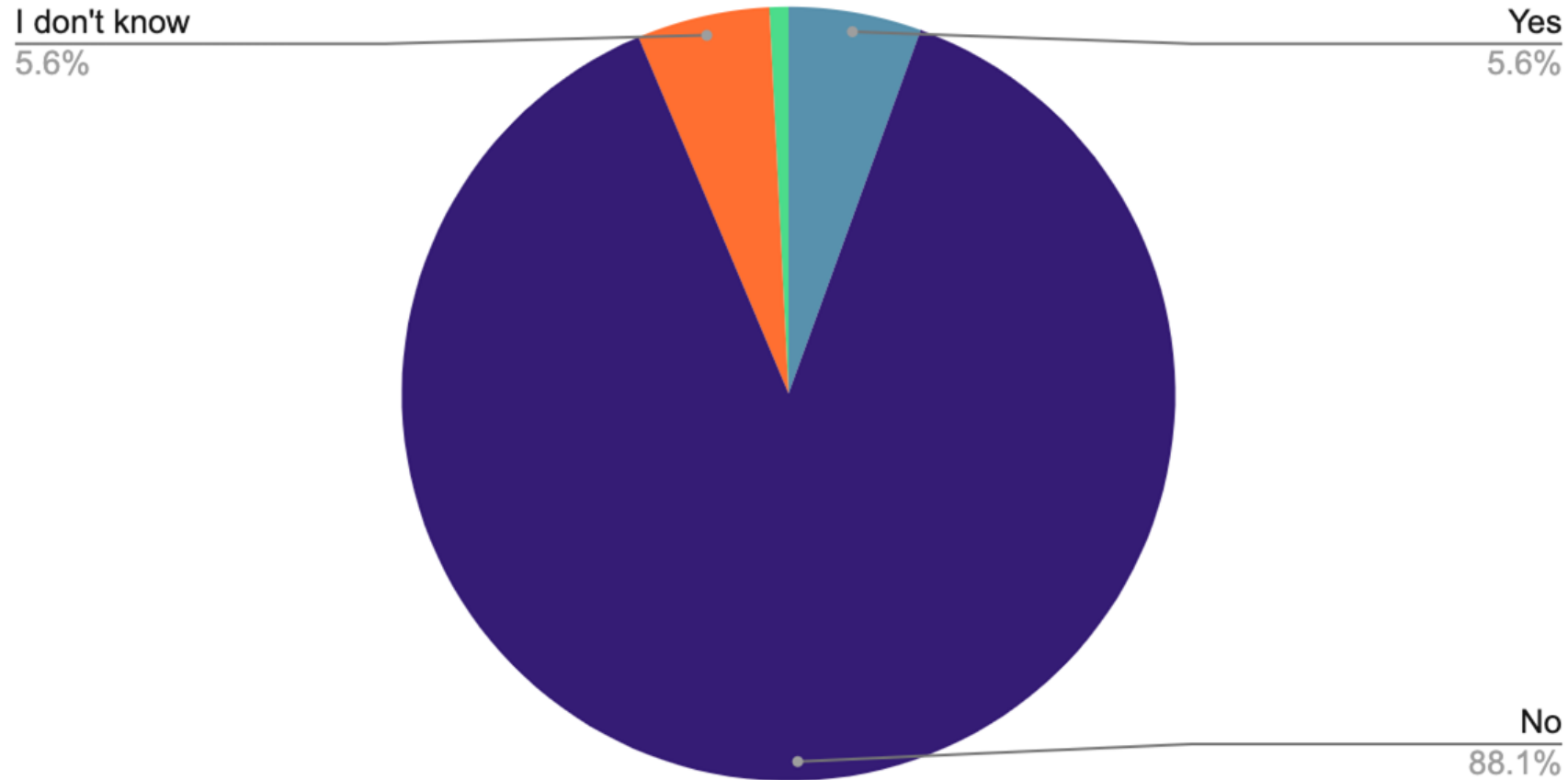
Does your child follow up with a dietitian?

I don't know
2.4%



No
97.6%

Does a specialist modify the formula amount for your child (based on Phe blood test results)?



International collaboration.. We all have a responsibility toward these children



