



HCU Network Australia

community for progress





Homocystinuria Patient & Caregiver Survey

Launched in June 2018

Aim:

Gain insight into subjective experience of patients/caregivers –

- Diagnosis
- Treatment
- Medical care

Participants:

Survey promoted via –

- our website, through emailing members, sharing on social media and a translated version was disseminated by Prof. Kozich.

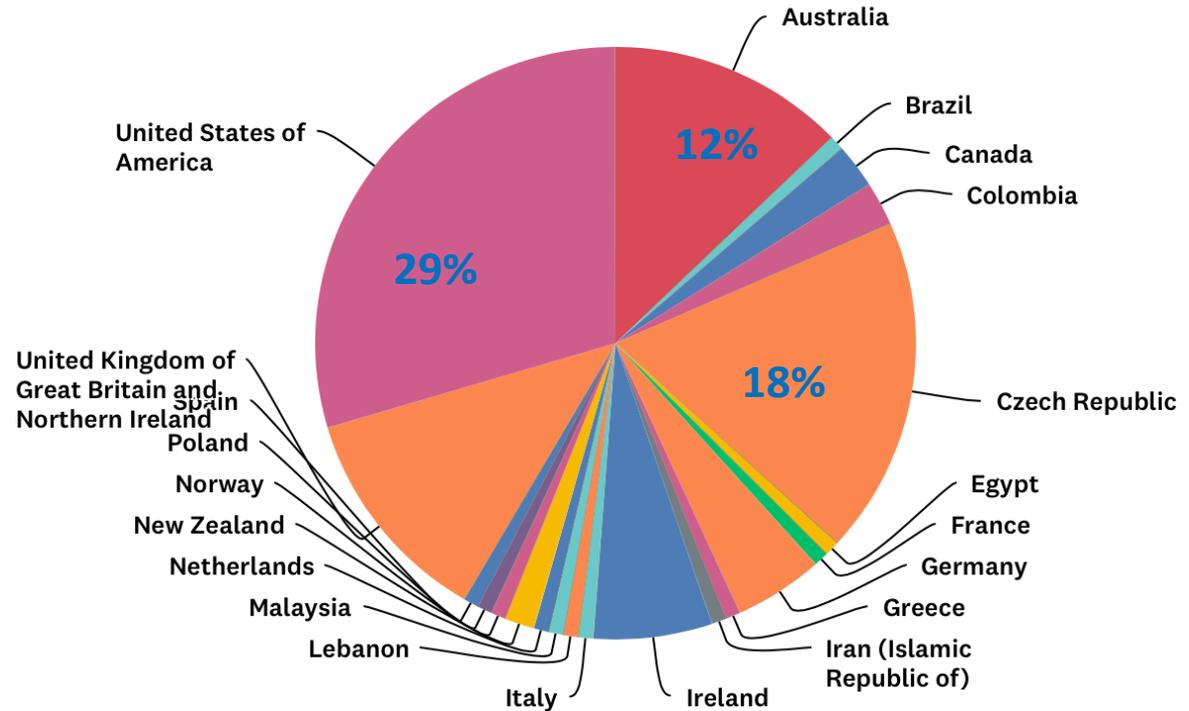
Acknowledgements

- Professor Viktor Kozich, Prague
- Margie McGlynn, HCU Network America.

Demographics

Data as of February 2019

- **128** respondents
 - **45** affected adults
 - **2** affected adolescents
 - **63** caregivers
 - **14** other
- Majority of respondents from **USA, Czech Republic and Australia**

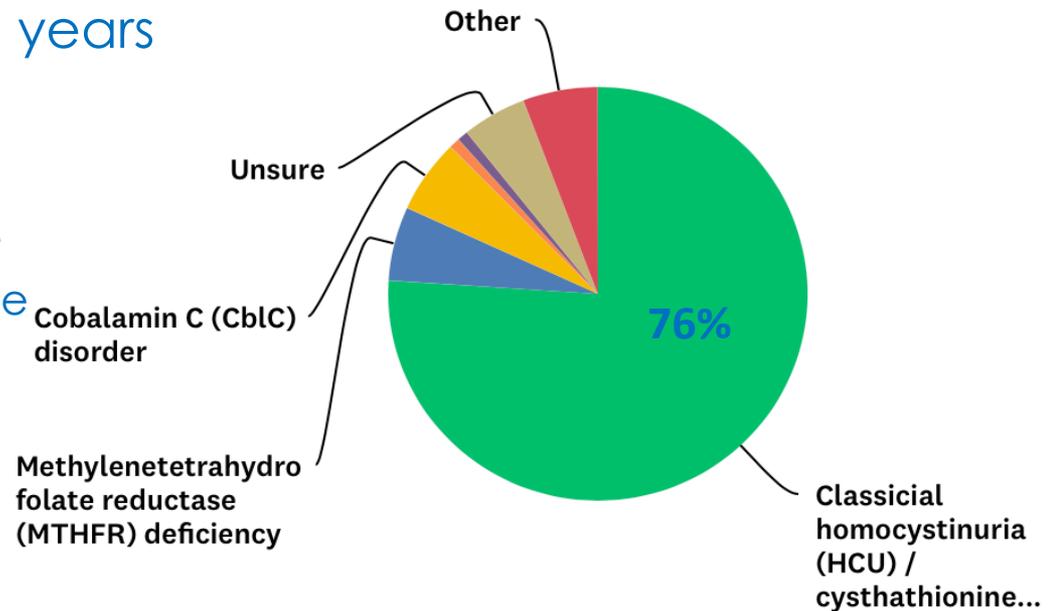


Country of residence



Age & disorder type

- Age range **0 – 65** years
- **92** CBS deficiency
 - 25 B6 responsive
 - 56 B6 non-responsive
 - 6 B6 partial responsive
 - 18 unsure
- **7** MTHFR deficiency
- **7** cbIC disorder
- **1** cbIE deficiency
- **1** cbIF disorder
- **6** unsure and **7** other



Diagnosis

To determine the **potential delays** and **impacts** on patients, respondents were asked:

- Whether detected by NBS
- Where they were incorrectly given another explanation for their symptoms
- The time take from presentation of symptoms to diagnosis
- The amount of doctors seen prior to reaching a diagnosis

Q Was newborn screening available at birth:

Yes **41** respondents

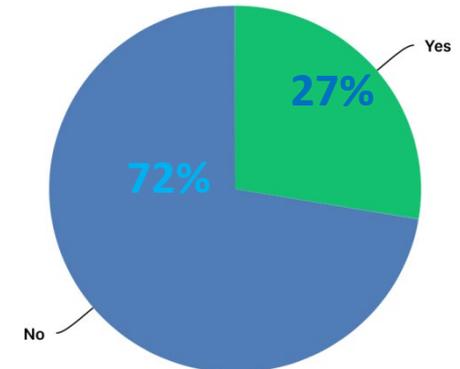
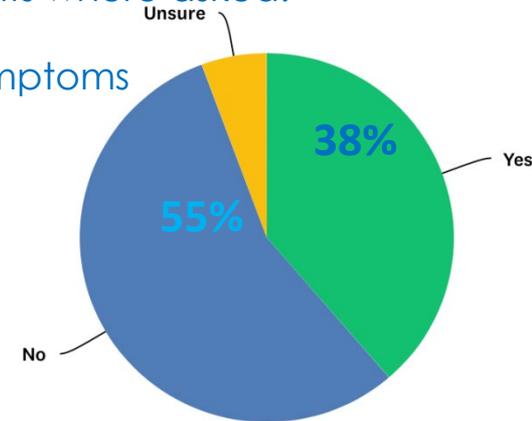
No **59** respondents

6 unsure

Q Were you diagnosed as a result of newborn screening

29 Yes

76 No

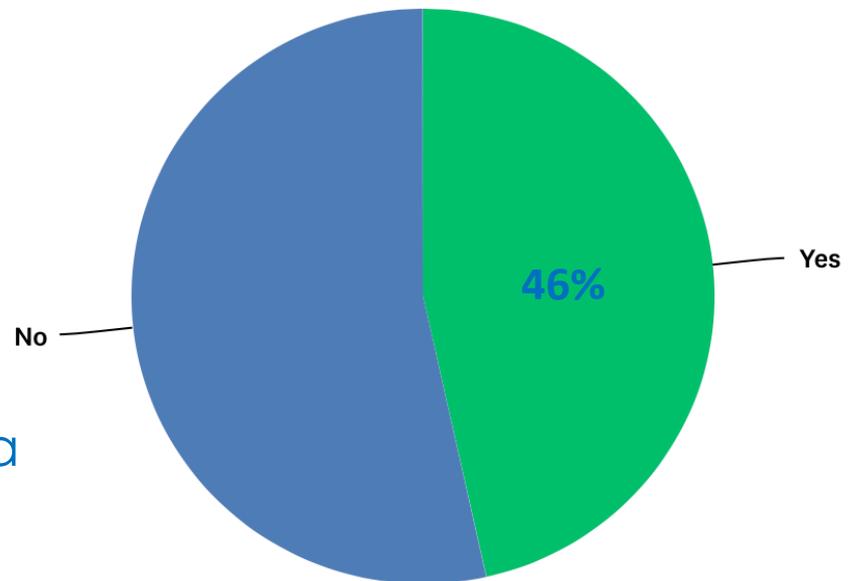


Diagnosis cont.

46% were incorrectly given another explanation for their symptom

Diagnosis:

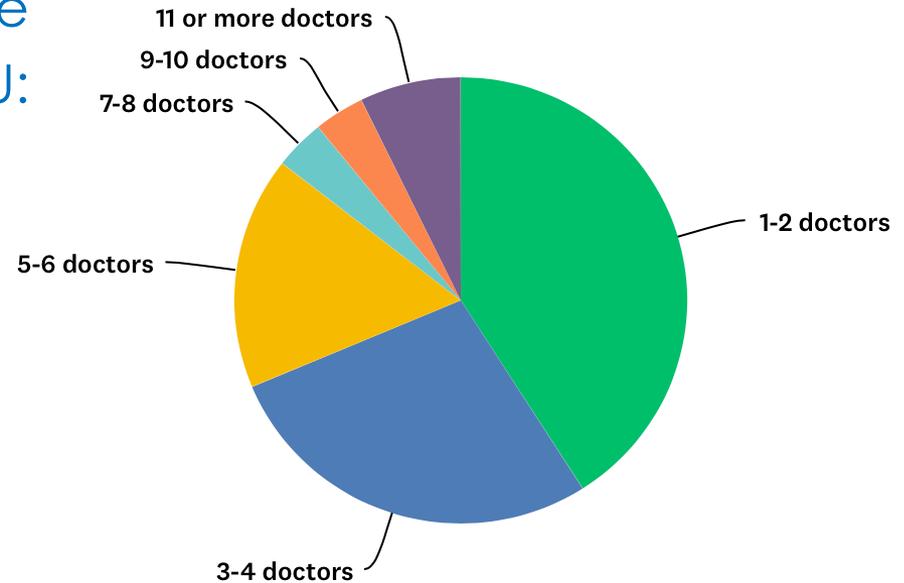
- Marfans
- Clinical depression
- Autoimmune disease
- Autism Spectrum Disorder
- Apraxia of speech & dyspraxia
- ADHD
- Developmental delay



Diagnosis cont.

Which specialist initially raised the Possibility of the diagnosis of HCU:

- Metabolic specialist
- Ophthalmologist
- Neurologist
- GP
- Geneticist
- Hematologist
- Endocrinologist
- Paediatrician



How many doctors seen before diagnosis
> 60% saw 3 or more doctors



Symptoms

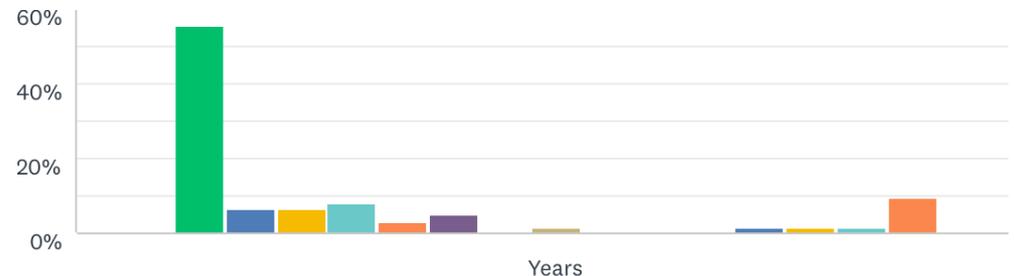
Symptoms prior to diagnosis:

- **Developmental delay** 23 respondents
- **Intellectual deficit** 8 respondents
- **Clumsiness** 13 respondents
- **Anxiety, seizures & depression** 6 respondents each
- **Obsessive compulsive disorder** 3 respondents
- **Behaviour disorder** 4 respondents
- **Learning difficulties** 11 respondents
- **Scoliosis & protruding/sunken chest** 5 respondents
- **Blood clots & strokes** 4 respondents
- **Pulmonary embolism** 2 respondents
- **Near sightedness & Lens dislocation** 12 respondents

Diagnostic delay

How long after first seeking medical advice for symptoms was a diagnosis of HCU discussed:

- Up to 1 years **55%**
- Up to 15 years **10%**



Perceived reasons for delay:

- **77%** Lack of knowledge about the disease amongst GPs
- **65%** Lack of knowledge about the disease amongst specialists
- **35%** Lack of symptom awareness by family
- **19%** Delays in obtaining test results
- **46%** Lack of access to appropriate tests
- **17%** Long waiting times to see medical specialists



Treatments

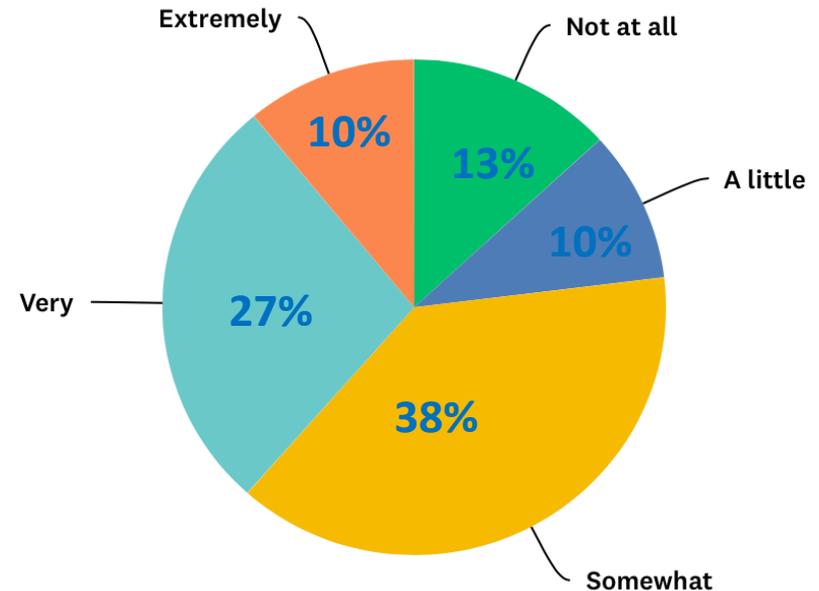
To determine the potential impacts of current treatments respondents were asked to report their feelings on treatment options.

Treatments identified:

- **70%** Low protein diet
- **55%** Amino acid mixture
- **75%** Betaine
- **22%** Intramuscular injections of vitamins
- **81%** Oral vitamins

Issues identified with formula:

- Arguing with child about taking formula
- Difficulty taking formula
- Difficulty eating our or doing something unplanned due to formula



Are you/your child satisfied with current treatment regime?



Amino acid mixture

15% reported **never** having difficulties taking formula
Remainder reported varying degrees of difficulty
70% reported having **missed** taking formula as directed

Open responses

“The taste is horrible and difficult to consume”

“He is 2 and we have to syringe him the formula. He often gags and every now and then vomits”

“Between the medicine and formula, it is a lot of strong tasting stuff”

“She is embarrassed to drink it outside of home”

“Refuse to drink it, taste is awful”

“A more palatable drink would make life easier”

Diet

How respondents felt about dietary restrictions:

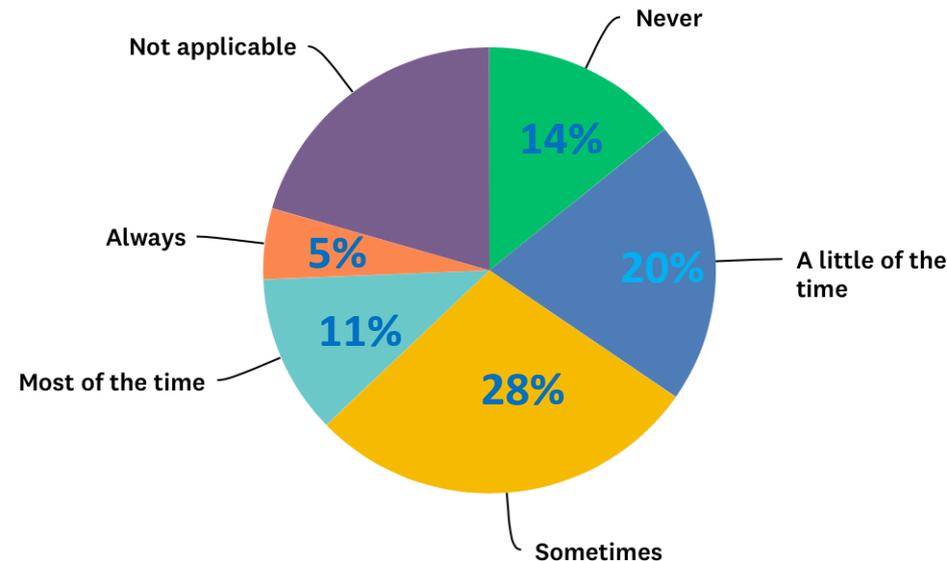
35% felt **unhappy** sometimes or most of the time when diet was restricted.

15% **did not** find it challenging to weigh/estimate protein.

67% wanted to eat **things others could**.

26% **did not** have access to low protein foods.

41% **always** followed the low protein diet as directed.



In the past 7 days was it hard for you/child to follow the low protein diet?





Use of health services

90% attended a metabolic clinic

37% rated their medical care as **excellent**

9% rated **fair** or **poor**

When contacting their clinic outside of appointments **42%**
always got an answer to their medical question.

10% **rarely** or **never** did

31% found it **very easy** to talk to medical staff about the disorder.

18% found it **difficult** to varying degrees

Summary

- **Medical awareness/education is lacking**
 - Requires attention to reduce the diagnostic odyssey of some individuals
- **Treatment options are necessary**
 - Requires attention to improve compliance and quality of life
- **Patients have access to quality healthcare services**

Survey remains open **30 April 2019**

HCUNetworkAustralia.org.au/experience-of-diagnosis-and-patient-satisfaction/

