QUICK GUIDE to
CLASSICAL HOMOCYSTINURIA
A treatable disorder

This **QUICK GUIDE** summarises key points from the information booklet: **CLASSICAL HOMOCYSTINURIA – A treatable disorder**. The booklet is designed to help patients, their families and caregivers understand more about classical homocystinuria (also known as cystathionine beta-synthase (CBS) deficiency).

Information in the booklet is based on the recently published Guidelines for the diagnosis and management of cystathionine beta-synthase deficiency[1] written as a guide for doctors and other specialists.

To view and download the full booklet, visit **www.hcunetworkaustralia.org.au**

Quick Reference: Booklet Page Number

What is CLASSICAL HOMOCYSTINURIA (HCU)?

• Classical homocystinuria is the most common of the homocystinuria group of disorders.	2
• HCU is a metabolic disorder where your body is unable to process an important amino acid called homocysteine correctly.	3
• In HCU, the CBS enzyme is faulty which affects the conversion of homocysteine to another important amino acid called cysteine; high levels of homocysteine begin to build up in your body which over time can lead to problems.	3
• If untreated, high levels of homocysteine can cause harmful symptoms that commonly affect the eyes, skeleton, brain and blood vessels.	4
• People with HCU can be divided into two groups: + Those who respond to vitamin B6 (pyridoxine) treatment + Those who do not respond to vitamin B6 (a more severe form of HCU)	4
• HCU in an inherited disorder; an affected child will have inherited two faulty copies of the CBS gene, one from each parent.	5
• HCU is a rare disease affecting about 1 in 100,00 people in Australia,[2] although it can be more common in other countries around the world.	6

How is HCU diagnosed?

• Because HCU is a rare disease with variable, non-specific symptoms it takes, on average, 4.5 years before a positive HCU diagnosis is made.[3] About half the cases of HCU will initially be misdiagnosed.	7
• Diagnosis is usually based on a combination of symptoms and biochemical tests.	7
• Newborn screening tests, where available, can help detect HCU in babies – but does not identify all HCU cases.	8
• Initial testing is confirmed by either enzyme or DNA analysis. Other members of the family will also be offered screening for HCU.	8
• Once HCU is confirmed you will be tested to see if you are responsive to vitamin B6 treatment.	8

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HCUNetworkAustralia.org.au

How is HCU treated?

Living life with HCU

Making decisions and finding support

References: 1. Morris A, Kožich V, Santra S, et al. J Inherit Metab Dis. 2017; 40:49–74. 2. HCU Network Australia, unpublished data. 3. HCU Network Australia, unpublished data.

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