

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 deficiency¹ 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)?

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| • 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 is 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?

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| • 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 |

How is HCU treated?

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| • The aim of HCU treatment is to keep homocysteine levels in the body as normal as possible while maintaining normal growth and nutrition. | 10 |
| • HCU treatment may include taking vitamin B6 and eating a diet low in natural proteins. | 10 |
| • Dietary management (low-protein diets with additional supplements) is recommended for everyone who does not respond to vitamin B6 treatment. | 10 |
| • Diets may need to be supplemented with vitamin B12 or folate – both are required in the metabolism of homocysteine back to methionine. | 11 |
| • Betaine may sometimes be added to the diet to lower homocysteine levels. | 12 |
| • Monitoring homocysteine (and other amino acids) levels and nutritional assessments play an important part of HCU treatment. | 12 |
| • Having surgery, being ill or travelling are examples of times when you may need to pay extra attention to HCU treatments. | 14 |

Living life with HCU

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| • HCU cannot be cured. Early diagnosis and treatment can help prevent complications occurring. If HCU is diagnosed late treatment can help prevent the progression of existing symptoms. | 15 |
| • Eye problems in untreated HCU are common and are often an early warning sign of HCU. | 15 |
| • Bone problems (how they form and grow) are also common in untreated HCU but are not usually seen in very young children. Older people with undiagnosed HCU can have facial features similar to those with Marfan syndrome. | 15 |
| • HCU affects the vascular system and if untreated increases the risk of blood clots forming – which can lead to strokes and other vascular problems. | 16 |
| • HCU is not thought to affect fertility. Contraceptives that contain oestrogen can increase the risk of blood clots and should be avoided. HCU also increases the risk of blood clots during pregnancy and after the birth; anticoagulant therapy may be recommended. | 17 |

Making decisions and finding support

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| • Ask your medical care team as many questions as you need to. Do not be afraid to ask them to explain anything you do not understand. | 18 |
| • Managing a metabolic disorder is a team effort – you will need support from your medical care team, family and friends. | 18 |
| • Find out as much as possible about HCU and its treatment, so you can talk over any problems or concerns you have with your medical care team. | 18 |
| • There are various on-line sites that can give you extra information on HCU including downloadable booklets designed especially for children. | 19 |

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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Disclaimer: This guide is for information only and should not be relied upon in place of medical advice. Any medical information is not intended as a substitute for informed medical advice. Consult a doctor or other health care professional for diagnosis and treatment of HCU. While all reasonable care in compiling the information has been made we make no warranty as to its accuracy.

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