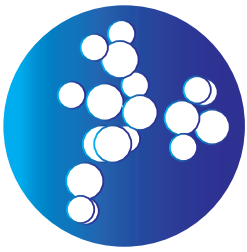
HOMOCYSTINURIA

WHAT IS HCU?



The Homocystinurias are a group of **inherited metabolic disorders** leading to accumulation of homocysteine and its metabolites in the blood and urine. **Classical Homocystinuria** (HCU) is the most common of these disorders*, caused by a deficiency in the enzyme known as **cystathionine beta-synthase** (CBS).

HOW IS HCU DIAGNOSED?

In Australia, Homocystinuria is screened by newborn screening but **screening does not detect all cases of Homocystinuria** and is not available in all countries. If not diagnosed by newborn screen, it can take an average of **4.5 years for an accurate diagnosis** of HCU[1]. Early diagnosis and **treatment** can make a **real difference** to patient outcomes[2].

WHAT ARE THE SYMPTOMS?

Ocular (Eyes)
- Severe and progressive nearsightness
- Lens dislocation

Central Nervous System
- Developmental delays and intellectual deficit
- Seizures
- Clumsiness
- Psychiatric disorders
- Behavioural problems

Skeletal
- Excessive growth of bones
- Protruding or sunken chest
- Highly arched foot
- Scoliosis
- Osteoporosis

Vascular
- Blood clots in the legs (deep venous thrombosis)
- Strokes (e.g. saggital sinus thrombosis)
- Blood clots in the lungs secondary to deep venous thrombosis (pulmonary embolism)

(mildly affected individuals may present as adults with blood clots as their only problem)

The **non-specific** nature of the signs and symptoms can lead to **under diagnosis**[3].

A TREATABLE DISORDER

No cure has been discovered for Homocystinuria. Treatment involves a strict low protein diet coupled with supplementation. There are two forms of treatment:

B6 responsive: B6-responsive patients respond well to high doses of vitamin B6 and most will also have folic acid supplementation. This may be all the treatment they need.

B6 non-responsive: B6 non-responsive patients require treatment involving a low protein diet, medical formula, betaine, folic acid and sometimes B12.

Information provided is for educational purposes only.
Please consult your doctor regarding any symptoms you might be experiencing.

For more information, visit HCUNetworkAustralia.org.au

HCU Network Australia
HCUNetworkAustralia.org.au

*The other forms of Homocystinuria are not discussed here.

[1] HCU Network Australia, unpublished data
[2] Schiff M, Blom HJ (2012), Treatment of inherited homocystinurias. Neuropediatrics 43, 295-304
[3] Garcia-Jimenez MC, Baldellou A, Garcia-Silva MT, et al. [Epidemiological study of the metabolic diseases with homocystinuria in Spain]. An Pediatr (Barc). 2012;76(3) 133-9