LIVING WITH HCU

facts, figures, and the importance of getting screened at birth

HCU Network America

This is Bill.

Bill suffers from a rare congenital disease called Homocystinuria, or

HCU

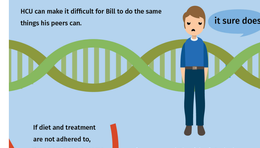
HCU expresses itself in different ways in different individuals, because of different genetic defects that cause the disease--

--and how well it is controlled via

diet & treatment

THE ORGANS AFFECTED INCLUDE

HCU can make it difficult for Bill to do the same things his peers can.



If diet and treatment are not adhered to, blood clots can become a threat.

Left unmanaged, the risk of stroke becomes high, especially in adults. Bill must eat foods high in folic acid and B-12, such as citrus fruits and plenty of vegetables, and take medication prescribed by his doctor.

Eyesight is one of the most affected abilities.

Many people with HCU are extremely nearsighted. Unfortunately, their vision often gets worse with time if not diagnosed or noncompliant with their treatment plan.

Bill also has to get his eyes checked regularly, because the lens of his eye can easily dislocate, which would cause blindness if left untreated.

Bone density and shape is also a concern.

Bill's arms and legs may be longer than his friends' limbs, as this trait occurs in some people with HCU.

He's also at an increased risk of bone loss and skeletal deformities such as scoliosis. It's important for him to have routine bone screenings, especially as he ages.

Less serious symptoms can include a flush across the cheeks, thin skin and...

...brittle hair.

DIAGNOSING HCU

Bill was diagnosed with HCU when he was born. He went through a screening process in the hospital, like all babies in the United States do.

A simple amino acid screen of blood and urine can shows elevated methionine levels, which indicates HCU.

Some HCU patients are missed in screening, so it's important for physicians to know the symptoms of HCU and test older patients.

HCU is inherited genetically, but since it's a recessive trait, it will only show up when both parents have a copy.

HCU is so rare because even when both parents are carriers, there is only a 25% chance the child will have the condition. There is a 50% chance they will only be carriers of the disease themselves. The remaining 25% will not have or carry HCU at all.

There are currently

1

in 200,000

people with HCU living in America.

LIVING WITH HCU

Because there is no cure for HCU, people with homocystinuria need life-long treatment. One of the most effective ways to manage HCU is through diet. Bill's body creates elevated levels of methionine and can't break down homocysteine, which means he needs to limit the amount of methionine he ingests. Methionine is an amino acid found in foods like chicken, fish, eggs, and milk.

Sticking to a diet with very little protein and lots of vegetables, like a vegan diet, is ideal.

However, patients should be mindful of what they are consuming. Even though vegetables and fruits are generally lower in methionine, there is some produce that has higher amounts like potatoes and corn. Moderation is key.

Taking supplements like folic acid, B 12 supplements, and vitamin B 6 are quite helpful for many patients.

Dietary formulas and betaine are also used by many people to successfully lessen their risk.

RISK FACTORS

Certain activities can be risky for people with HCU, making a blood clot, stroke, or other medical event more likely.

Pregnancy

Long periods of inactivity, like on airplanes.

Estrogen-based medications like birth control

Dehydration

Not sticking to the diet or treatments.

For people with HCU, compliance to their doctor's orders is very important. Becoming active in the community can help patients adjust to their new lifestyles. To learn more about HCU and access helpful resources, please visit us at HCUNetworkAmerica.org.

www.hcunetworkamerica.org

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