# Classical Homocystinuria

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Hello! My name is Henry. I have Classical Homocystinuria, and I really like the color blue.

For me, blue means the sky and sea, the mountains, and my dreams. Maybe that's why I love watching birds...

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Let me tell you my story, and the story of my dreams.

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I was born a healthy baby from a very friendly family and soon I left the hospital.

My parents told me that I was breastfed and I started to walk early.

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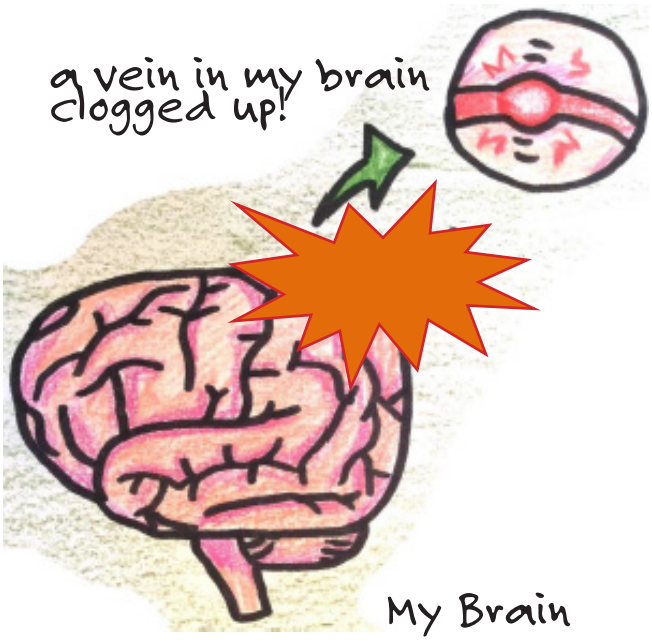
But, when I was almost 2 years old, I had a problem: all of a sudden, a vein in my brain clogged up! Can you believe that?

Because of this, I fainted and had a seizure, and my parents rushed me to the hospital.



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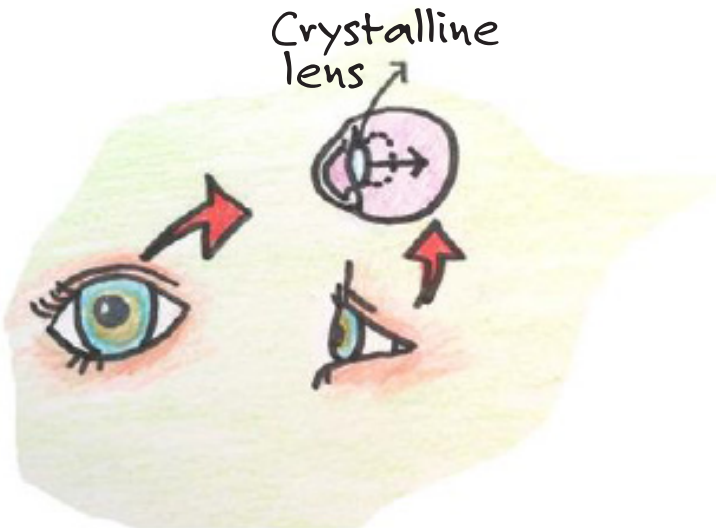
The whole family got very anxious, but the hospital staff treated us very well and soon I was fine. While I was recovering, a medical geneticist came to see me. And an eye doctor too...

She said I also had a problem called "dislocation of the crystalline lens"... The crystalline lens is a lens within our eyes... this lens has to "stand up straight" in the eyes and, when it moves out of place, you have a dislocated lens...



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I didn't understand it then, I was too little, but I found it odd. Geneticist was also a very strange word... My parents, when I grew older, explained to me that a medical geneticist is a doctor who cares for people with rare diseases.

It is rare for a child to have clogged veins in the brain and an eye's lens out of place, that's why she came to see me, I think.

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Then, they did blood tests on me... And soon they found out that I had a rare genetic disorder called Classical Homocystinuria.

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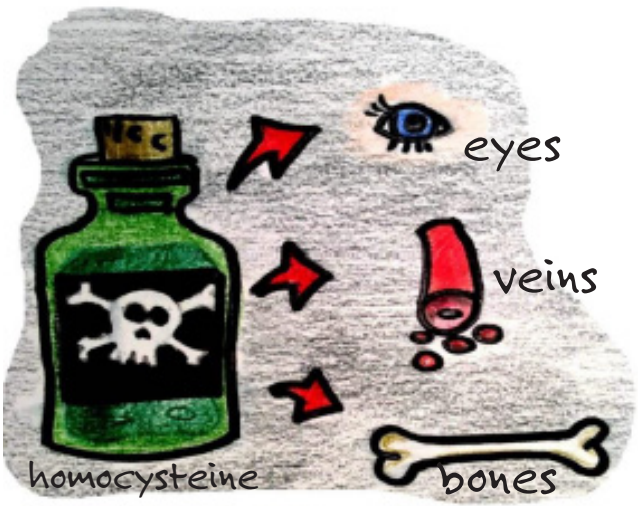
People who have Classical Homocystinuria produce in their body very small amounts of a substance called cystathionine beta-synthase (it's easier to just call it CBS). When the CBS is not working right, there is a buildup of homocysteine in the blood.

When blood homocysteine levels are high, homocysteine acts as a poison in our bodies: it causes the veins in the heart, brain, or legs, for example, to clog; it causes the lens of the eye to move out of place (that's why I wear glasses and had an eye surgery); it makes me more likely to have osteoporosis ("weak bones")...



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But Classical Homocystinuria is treatable... There are several vitamins (which I take) that help lower homocysteine levels in the blood...

Some people with Classical Homocystinuria can lower their homocysteine levels when treated with just vitamins (that's not my case). I also need to eat a special diet, drink a special milk and take special drugs... I'm used to the diet, but I have to watch out to always stick to my diet...

Homocysteine is produced from amino acids, which are the "building blocks" of proteins (proteins are found in meat, for example), and, because of that, I have an almost vegetarian diet... Very healthy!

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I will need treatment for the rest of my life.... After I started my treatment, I didn't have any other problems and I plan to keep without problems.... I know it depends on me to continue with treatment, visit the doctors who care for me, see the dietician, have blood tests to check my homocysteine levels... And to have dreams...

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What are my dreams? Well, I'm studying to get into college. But my biggest dream is that people learn and research more and more about Classical Homocystinuria…

I wonder if one day there will be a cure for my problem...who knows?

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Sometimes I also wonder... is Classical Homocystinuria really rare or are there lots of people like me in the world? But, unlike me, they just don't know they have Homocystinuria...

This is dangerous: if we don't know we have Homocystinuria, we can't receive the right treatment...

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Do you want to be part of our family and help me make my dreams come true?

If yes, remember that some specific signs strongly suggest that a person, even if being a child, a teenager or an adult, have Classical Homocystinuria (see next page). These people need to be evaluated by a specialist to have their diagnosis confirmed as early as possible and to receive appropriate treatment.



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Signs suggestive of Classical Homocystinuria:

High homocysteine levels in the blood.

A dislocation of the eye's lens. The eye's lens is called crystalline lens.

Clogged veins anywhere in the body, specially in children and in teenagers. Doctors call clogged veins "thrombosis".

Newborn screening test showing high levels of methionine in the blood.

Presence of relatives with Classical Homocystinuria, especially siblings. Parents who have a child (boy or girl) with Homocystinuria have a 1 in 4 (25%) chance to have another child (boy or girl) with the same problem. That's why genetic counseling is so important!!!

Henry



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This publication is intended for patients with Classical Homocystinuria, their families and friends.
If you have any questions about this disease, contact :

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