

explainity explains: Cleidocranial Dysplasia

This is Maria and her son Paul. He was born with a hereditary condition called cleidocranial dysplasia. When he was born, Maria was quite worried about this. But don't stress, Paul is doing well today. So What effects does this hereditary disease have on him?

“Dysplasia” stems from a Greek word and means malformation. “Cleido” refers to the clavicles or collarbones, which Paul is missing. This is why he has highly flexible shoulders. “Cranial” stands for the skull. Right after birth the skull plates of a newborn are slightly malleable and solidify after some months.

But Paul's plates have never completely closed this gap. As a precaution he wears a helmet when he does sports or rides a bicycle.

In addition, Paul has a small jaw and his baby teeth did not want to fall out for a long time. The doctor had to remove them instead. Later on, he had more second teeth than usual. Because his jaw is still too small, these teeth don't have enough room and have to be corrected. The most common form of treatment for kids with cleidocranial dysplasia is surgery on the jaw.

These symptoms are caused by Paul's genes. They have a mutation on the RUNX2 gene. One in one million people worldwide are affected by this mutation. If one parent carries the mutated gene, there is a 50% chance of them passing it on to their offspring. Even healthy parents can have a child with cleidocranial dysplasia, if the mutation happens spontaneously.

Paul's bone structure and growth are also affected. The density of his bones has to be monitored by a doctor regularly. Otherwise he might have fragile bones later in life. Still: With a little help, Paul can lead a long and happy life and do anything he wants to do.

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