



Hi folks, my name is Mara, I'm pretty accurate 1.5 years and the princess of the family. I'd like you to tell my story. Shortly after I was born, my mom had feel like I'm a little different. She is a nurse for healing education and probably has a good eye for it. At first, my mom was called crazy. Doctors often told her I wasn't sick. My mom should get some therapeutic help. Anyway, I couldn't drink well from the start, didn't put on weight well. I still don't do that and keep my slim 8 kilos at 74 cm height for several months. The doctors always said everything was okay. But Mama didn't give up. I didn't develop well and after 7 months it was so bad that the doctors were start serious wondering what I might have. They searched for a long time. And now my diagnosis finally came to the light. A genetic defect on TRAPPC11 causes me to develop an LGMDS2. So far, I'm just lame. I can't turn around and Mom says my arms are like pudding. I don't know what's wrong with that. Pudding is delicious. I can even eat it. I can't eat anything that isn't mashed food.



Otherwise, except for muscles, I don't miss anything about me. The genetic defect gives me a bunch of other unpleasant effects. I have very bad eyes (that was the first thing I noticed) also my head is too small (microcephaly) and some things are not right in my head (beam hypoplasia and more). Anything that my friends of the same age than me can do, I can't. I can't crawl or sit. It's hard for me to turn around myself. When I grip my toys, the hit rate isn't that high. However, I am usually quite happy and in a good mood. My favorite person is my big brother Jannik. He's just the best. Well, my many construction sites make Mom very scared for me. She says my prognosis may not be so good, and she's afraid I might die early. She cries a lot and often. But then I smile as wide as I can, and then the world is a little better for my mom. I'm tough and I give everything.