GLUT 1 Deficiency

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GLUT1 Deficiency
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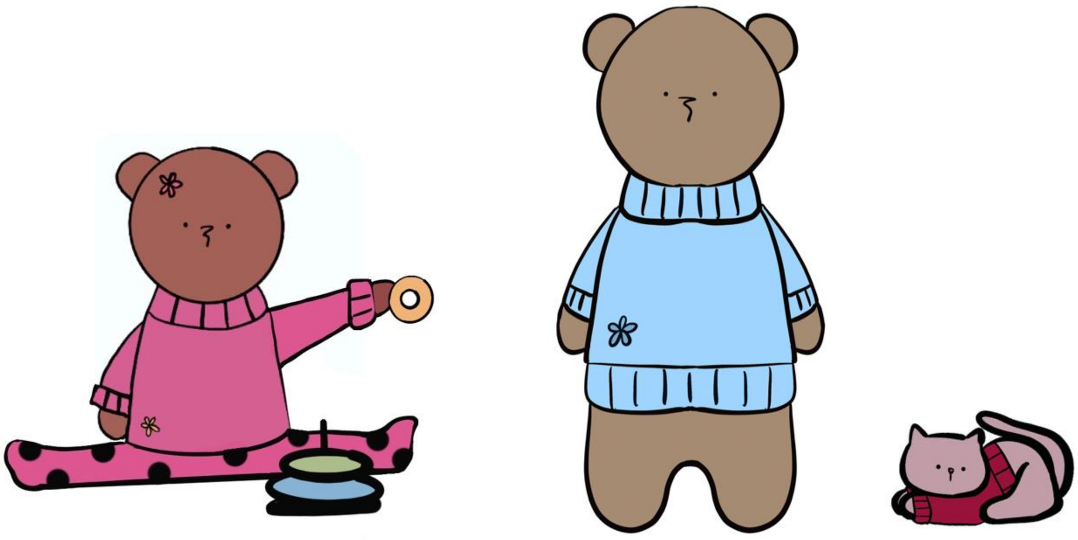
Hello! This is my little sister; we love to play together.

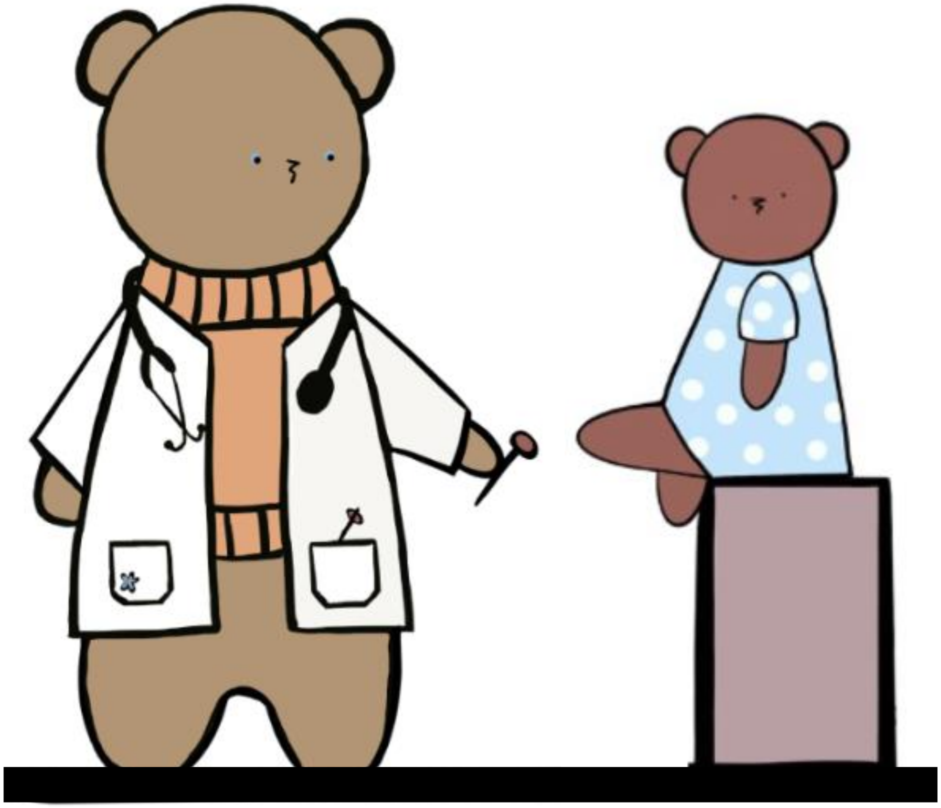
"Hello"



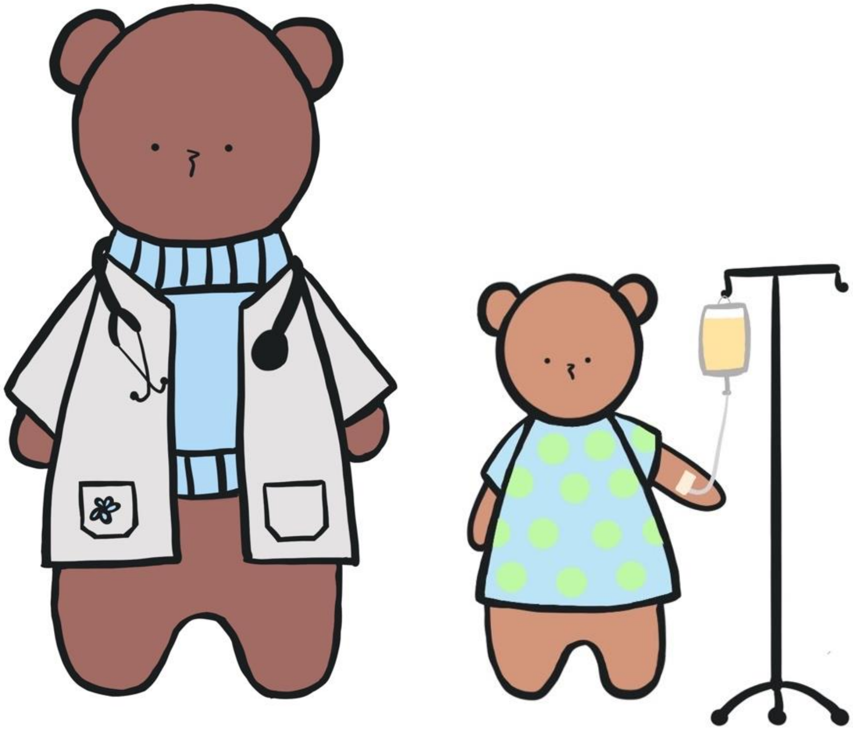
One day she suddenly started staring and shaking. My mom called 911.

I could tell that my parents were really scared. I was really scared too!

At the hospital, a very nice doctor explained that the shaking episodes were "seizures".



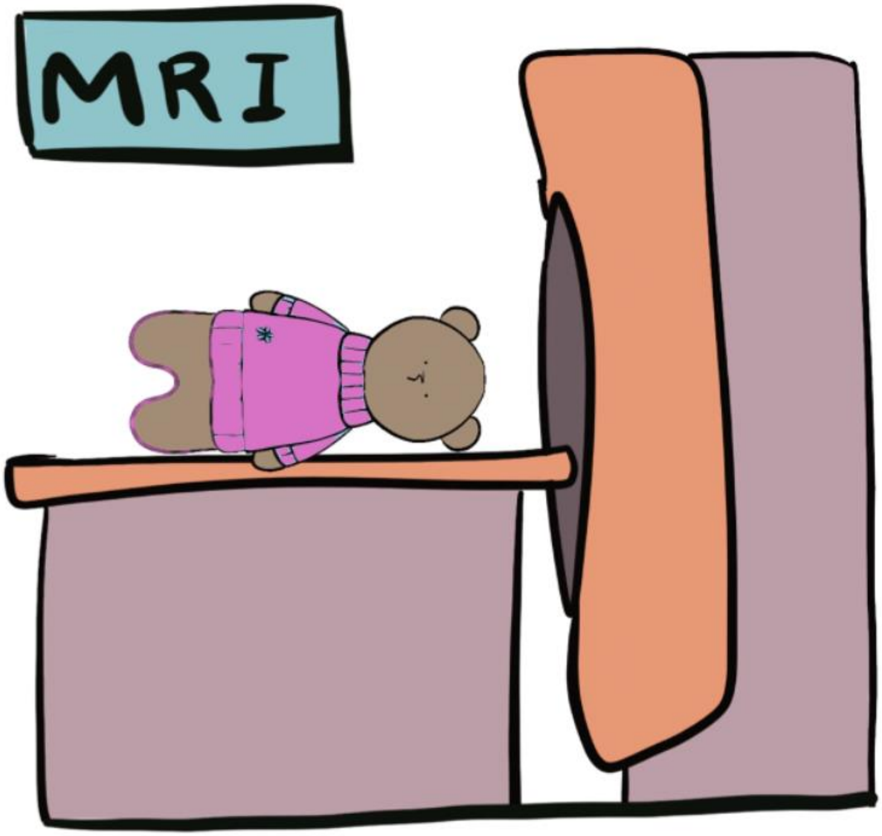
The doctor told my parents that she needed to stay at the hospital for tests.



She got a a test called electroencephalogram (EEG). It did not hurt.

During the EEG they put several stickers with wires in her head. There was also some flashing lights. All she had to do was to rest quietly and blow on a pinwheel.

The other test was called MRI, which took a picture of her brain. It did not hurt.

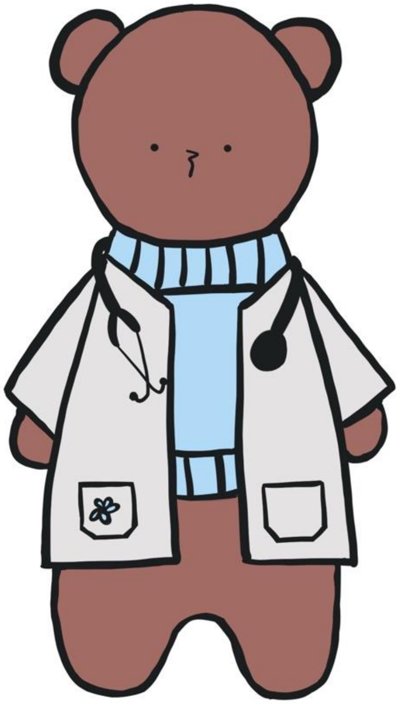


When she came back home, I was a little sad because my sister was not as playful as before.

The doctor told us that once the test results were done, they would be able to find the best way to help my sister.

During the follow up clinic visit the doctor said: Good news! We know how to help your sister.

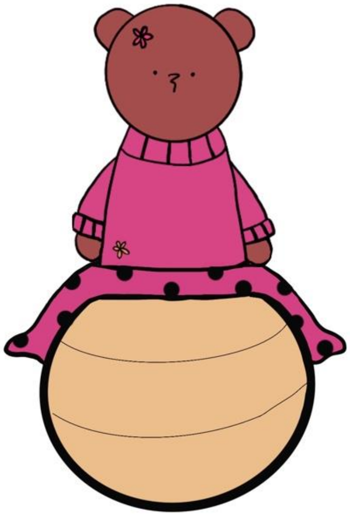
At first my parents were very scared, but they were happy to hear that doctors can help her get better.



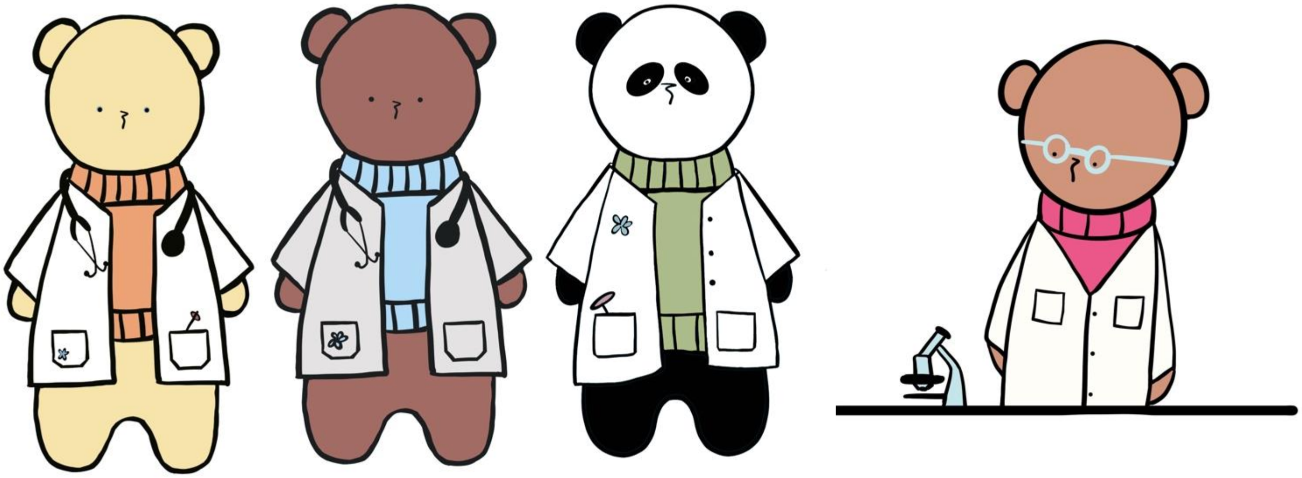
Later the doctor explained that my sister has GLUT1 Deficiency Syndrome.

- We are born with GLUT 1 deficiency, so you cannot "catch it."
- It is a genetic disease, a problem affecting a gene called SLC2A1. This is not anyone's fault.
- In addition to medication, the best treatment option is the ketogenic diet.
- Kids with GLUT1 deficiency cannot use glucose (sugar) as the brain main fuel. The Ketogenic Diet provides fat as the main fuel for the brain.

- A dietitian will help our family with the ketogenic diet.
- She even showed us a Ketogenic Diet recipe book that had a lot of different types of food. Even keto pizza, avocado chips, bacon and ice cream!
- Physical, occupational and speech therapy are also very important to help my sister learn new skills.

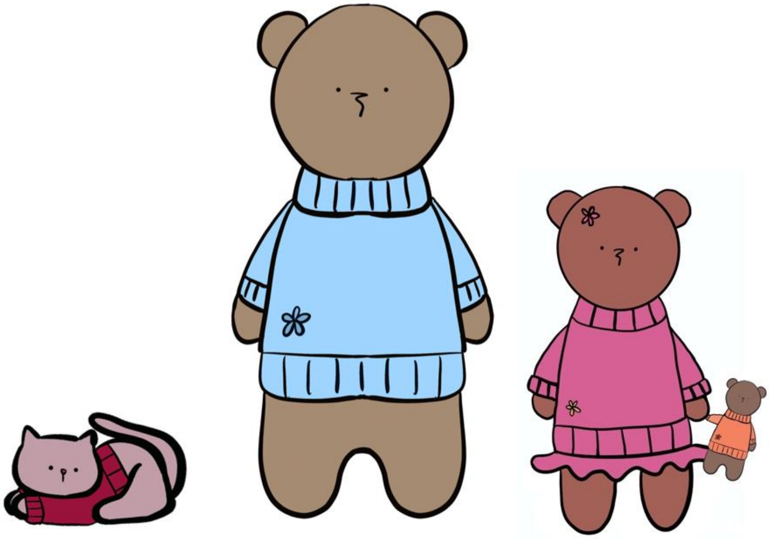


There is still so much to learn about GLUT1 deficiency syndrome and scientists are working hard to find out how to treat it even better.



In the beginning, I thought my sister was the only kid with GLUT1 deficiency syndrome, but now I know that many children also have it.

Everyone is a little different. Although many children have GLUT1 deficiency syndrome; they are still regular kids just like you and me.



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