



# Áine

## LIVING WITH AUTOSOMNAL RECESSIVE INFANTILE OSTEOPETROSIS

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Áine Nolan is almost 2 years old. In March 2021, just a year ago, she was diagnosed with a rare metabolic bone disorder called Autosomal Recessive Infantile Osteopetrosis.

Osteopetrosis, or stone bone, is characterized by overly dense bones throughout the body. Infantile Osteopetrosis affects 1 in 250,000 people and the only treatment is a bone marrow transplant as long as it is caught in the early stages.

Thankfully Áine was eligible for a bone marrow transplant. Since diagnosis she has had a lot of on-going health issues. She had a shunt fitted due to hydrocephalus and visual impairment due to damage done by her thickened skull bones. She has had numerous blood and platelet transfusions, Sepsis twice and has ended up in ICU twice.

Áine had a life-saving bone

marrow transplant on July 8th 2021 and is now doing so well after having a lot of complications in the early stages post-transplant. This includes numerous infections, graft versus host disease, haemolytic anaemia and mucositis.

Áine spent 108 days in isolation on a high dependency transplant ward in St John's, from June 27<sup>th</sup> to October 13<sup>th</sup> 2021.

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We, Áine's parents struggled at the start of her journey as she was always in and out of A&E and the doctor's office. We found ourselves explaining our daughter's situation to doctors and nurses because it is just such a rare condition. It wasn't something we were ever taught about.

Since entering the doors of St John's that feeling has definitely changed. They had prior knowledge of the condition as another patient had the same condition a few years previously. It was nice not having to explain everything from the very start again and again.

The support we have all been shown is absolutely incredible.....