‘Miracle’ drug grows young girls limbs, costs of rare disease treatments pose challenges

Evie Elsaesser is 5 years old and she loves to run. And that is a medical miracle.

When Evie was born, much of her skeleton remained soft and un-calcified. Her arms were short, her ribs tiny, and her legs bowed. They curled up under her as if she was doing yoga.

After two weeks the seizures started: Evie would stop breathing for minutes at a time. Her parents rushed her to the hospital, where a round of blood tests finally led to a diagnosis. Evie’s levels of alkaline phosphatase, a chemical important for bone hardening, were low. That meant she had an incredibly rare genetic disease called hypophosphatasia (HPP). The severe form afflicts one in 100,000 newborns.

The geneticist would later tell Lindsey he’d expected Evie to die within five months. Doctors could give her a medicine to help with the seizures, but there was no treatment for her weak skeleton. But then she was enrolled in a clinical trial for asfotase alfa, a drug being developed by Enobia Pharmaceuticals, a Montreal biotech startup with just two-dozen employees. The drug replaced the bone-forming enzyme she was missing.

Thanks to an experimental drug, a little girl who would have almost certainly died is running and laughing. This is exactly what we hope new medicines will do – truly change lives. Now for the question that determines how many such drugs we get: society, what is this worth to you?

Evie’s drug is likely to cost more than $200,000 per patient per year. The wholesale cost of other drugs to treat rare diseases can cost upwards of $500,000 a year.

Read full, original article: A Drug Regrew A Little Girl’s Missing Bones. How Much Should That Cost?