# Can gene therapy cure fatal diseases in children?

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In order for the body to function, a balance is necessary between the cells that build up the bones in our skeletons and the cells that break them down. In the disease malignant infantile osteopetrosis, MIOP, the cells that break down the bone tissue do not function as they should, resulting in the skeleton not having sufficient cavities for bone-marrow and nerves.

"Optic and auditory nerves are compressed, causing blindness and deafness in these children. Finally the bone marrow ceases to function and, without treatment, the child dies of anaemia and infections", explains Carmen Flores Bjurström. She has just completed a thesis which presents some of the research at the division for Molecular Medicine and Gene Therapy in Lund.

The researchers' work focuses on finding alternatives to the only treatment currently available against MIOP, namely a bone-marrow transplant. This treatment can be effective, but it is both risky and dependent on finding a suitable donor.

Gene therapy requires no donor, as stem cells are taken from the patients themselves. Once the cells' non-functioning gene has been replaced with a healthy copy of itself, the stem cells are put back into the patient.

Great hopes have been placed on gene therapy as a treatment method but the work has proven to be more difficult than expected. The method is used today for certain immunodeficiency diseases, and has also been

applied to a blood disorder called thalassemia.

"So far, the method is not risk-free. Since it is impossible to control where the introduced gene ends up, there is a certain risk of it ending up in the wrong place and giving rise to leukaemia. This is why gene therapy is only used for serious diseases for which there is no good treatment", says Carmen Flores Bjurström.

The Lund researchers have conducted experiments with gene therapy in both patient cells and laboratory animals. The next step is to conduct trials on patients. The trials will probably take place at the hospital in Ulm, Germany, which currently treats the majority of children in Europe suffering from MIOP.

MIOP is a rare disease: in Sweden a child is born with the condition approximately once every three years. Worldwide, the incidence of the disease is one case for every 300 000 births. It is, however, more common in Costa Rica where 3-4 children per 100 000 births have the disease.

"But there are several other genetic mutations that lead to other osteopetrosis diseases. If we manage to treat MIOP, it may become possible to treat these other conditions as well", hopes Carmen Flores Bjurström along with her supervisor, Professor Johan Richter.

**More information:** The thesis is entitled "Targeting the hematopoietic stem cell to correct osteopetrosis" and it will be presented on 6 September.

Provided by Lund University