

Gene-therapy success for children born without functioning immune system

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Children playing. Credit: Fiona Pragoff/Wellcome Images

(Medical Xpress) -- Researchers at the UCL Institute of Child Health have developed gene-therapy programmes that can successfully treat children born with an inability to fight infections, according to studies published today in *Science Translational Medicine*.

The therapies have been developed to treat two conditions commonly known as 'boy in the bubble' syndrome: X-linked severe combined immunodeficiency (X-SCID) and adenosine deaminase-deficient severe combined immunodeficiency (ADA-SCID). In both conditions the infant is born with an [immune system](#) so compromised that it is considered absent. [Children](#) have to live in a sterile environment - the so-called 'bubble'.

X-SCID is caused by mutations in the IL2RG gene, which governs the behaviour of a protein involved in the development of a number of immune-system cells. ADA-SCID results from the lack of an enzyme

that helps cells to get rid of toxic by-products.

The studies report that 14 out of 16 patients across the two programmes were successfully treated. The therapies were developed by Professors Adrian Thrasher and Bobby Gaspar of the UCL Institute of Child Health with funding from the Wellcome Trust, Great Ormond Street Hospital Children's Charity and the National Institute for [Health Research](#) Biomedical Research Centre at Great Ormond Street Hospital,

In the treatment of X-SCID, all ten children showed clear clinical improvement and were able to attend school, parties and nurseries, etc. One developed [leukaemia](#) and is currently in [remission](#). As a result, a safer method of implementing the therapy was developed and is now being used.

In a programme looking at ADA-SCID, four out of six children treated showed clear clinical improvement and, as with the X-SCID children, were able to socialise with other children in normal situations.

Children involved in both programmes continue on some medication, but five of the ten X-SCID and three of the four ADA-SCID patients no longer take immunogloblins.

Professor Thrasher, consultant in paediatric immunology and X-SCID programme lead, said: "These are excellent results for our [gene therapy](#) programmes and the first time we have been in a position to say we have found a cure for patients with these conditions. It demonstrates that gene therapy for immune diseases is now mainstream, and we hope this approach will benefit many more of our patients in the future."

Professor Gaspar, consultant in paediatric immunology and ADA-SCID programme lead, said: "We are delighted by the results of both of these programmes. The success of the ADA-SCID programme has also saved

the NHS millions of pounds, as these children are able to stop receiving regular costly enzyme replacement injections."

Worldwide, 30 children have received gene therapy for ADA-SCID; 70 per cent of them have seen a clear clinical benefit and none has died.

"We are now leading trials for two other immune disorders with colleagues from across the globe, and hope that gene therapy will now be seen as a standard alternative to conventional treatments such as bone marrow transplant," adds Professor Gaspar. "We also hope to be able to extend this approach to other conditions, such as lysosomal storage diseases, where a metabolic fault means chemicals build up in cells and cause various problems throughout the body."

Guy Harnden, now six years old, was enrolled on the X-SCID programme. His mother, Gaynor Harnden, said: "Guy is now doing brilliantly, he can do all of the things his friends can and more. He is able to play football and ride a pony.

"He wouldn't be here if it wasn't for the option of gene therapy treatment. We are incredibly grateful to the whole team at Great Ormond Street Hospital, but especially Adrian Thrasher and Bobby Gaspar who pioneered this work. To other parents who find themselves in our situation we would say 'go for it'."

Great Ormond Street Hospital runs more gene therapy trials for immune deficiency in children than any other centre in the world. The papers come over a decade after Rhys Evans became the first boy to be successfully treated for X-SCID by gene therapy at the hospital. The first patient to undergo ADA-SCID treatment is now almost eight years post gene therapy.

More information: Gaspar HB et al. Long term persistence of a

polyclonal T cell repertoire after gene therapy for X-linked severe combined immunodeficiency. Sci Transl Med 2011 [epub].

Gaspar HB et al. Hematopoietic stem cell gene therapy for adenosine deaminase-deficient severe combined immunodeficiency leads to long-term immunological recovery and metabolic correction. Sci Transl Med 2011 [epub].

Provided by Wellcome Trust

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