

Researchers finds better ways to improve the chances of survival of children with a rare immune deficiency

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An international study published in the journal *Blood* by researchers led by Dr. Elie Haddad, a pediatric immunologist and researcher at CHU Sainte-Justine and professor at Université de Montréal, highlights the urgent need to develop better treatment strategies for patients suffering from severe combined immune deficiency (SCID).

This deficiency, better known as "bubble baby disease" is a rare syndrome characterized by a total non-function of the body's immune system. The children affected have no [immune defence](#) and are vulnerable to bacteria, viruses and fungi, resulting in repeated severe infections. Without appropriate treatment, in most cases this disease is fatal within the first months after birth.

SCID can be caused by mutations in various genes involved in the functioning of the immune system. The new research shows that the nature of the mutated gene (or genotype) has a significant influence on patients' survival and reconstitution of their immune system after bone marrow [transplant](#). According to the study, the genotype must be taken into account when adapting [treatment strategies](#) to individual patients.

44 Medical Centres Involved

Funded by the U.S. National Institutes of Health, the Primary Immune Deficiency Treatment Consortium (PIDTC) involves 44 medical centres

across North America. The PIDTC conducted a retrospective analysis of 662 patients with SCID who received hematopoietic stem cell transplants as their first therapy between 1982 and 2012 in 33 of these centres, forming the basis of the new study.

"Immune diseases are among the top priorities at CHU Sainte-Justine in terms of care, teaching and research," Haddad said. "There are only one or two cases of SCID per year in Quebec. Having access to such a cohort of patients with this rare disease and over such a long period has provided us with significant and unique data to advance our knowledge in this area."

The results showed that survival rates of patients were higher after a cell transplant from a matched sibling donor. In recipients from other donor types, which represent 86 per cent of cases, the data showed that the SCID genotype had a strong influence on survival and [immune reconstitution](#). In addition, the researchers found that young age and absence of active infection at the time of transplant were also key factors for survival, and both were significantly associated with improved survival following a transplant.

Critical Need for Neonatal Screening

"We need to develop patient-specific treatment strategies," Haddad said. "There is a critical need for neonatal screening to establish appropriate isolation, implement infection-prevention measures, particularly before transplantation, and ensure rapid referral for [bone-marrow transplant](#) or gene therapy after diagnosis."

The study also points to the need for close monitoring of immune-system reconstitution after treatment to identify patients with conditions that may require additional intervention and prevent a poor long-term prognosis. Further studies will be needed to identify patient- and

transplant-related factors that limit early immune-system reconstitution, and to determine the most appropriate and effective interventions.

"In our future research, our objective will be to analyze the late effects of conditioning and the quality of long-term immune-system reconstitution after hematopoietic cell transplant for this deficiency," said Haddad.

"SCID genotype and 6-month post-transplant CD4 count predict survival and immune recovery," by Elie Haddad, H  l  ne Decaluwe, Pierre Teira et al., was published in the journal *Blood* on Oct. 25, 2018.

More information: Elie Haddad et al, SCID genotype and 6-month posttransplant CD4 count predict survival and immune recovery, *Blood* (2018). [DOI: 10.1182/blood-2018-03-840702](https://doi.org/10.1182/blood-2018-03-840702)

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