Early diagnosis can save babies' lives: A guide to severe combined immunodeficiency disease (SCID)

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A new review provides guidance on a deadly, but rare, disease that is potentially curable if identified early. Severe combined immunodeficiency disease (SCID), known as the "bubble boy disease" in the 1970s, is treatable with a stem cell transplant, gene therapy and other treatments if identified at birth or soon after.

The review, published in *CMAJ (Canadian Medical Association Journal)*, is aimed at pediatricians, family physicians and other doctors who may treat newborns, including those who appear healthy at birth but begin to get severe, repeated infections requiring emergency department visits.

The death rate for severe combined immunodeficiency disease is at least 30%, with infection causing 60% of deaths in infants.

Ontario was the first jurisdiction in Canada to offer screening for severe combined immunodeficiency disease in 2013 as part of the heel prick test performed soon after birth. Screening has been expanded to the Maritime provinces and will be implemented in several other provinces.

"This review informs physicians who may treat newborns in their practice (e.g., family physicians, obstetricians, pediatricians) on how to approach patients and counsel families who are faced with an abnormal screen," writes Dr. Stuart Turvey, a professor with the UBC Department of Pediatrics and an investigator, BC Children's Hospital, Vancouver,

BC, with coauthors.

The review includes features to watch for in newborns with repeated illnesses, information on screening and diagnosis, and treatments for the disease, as well as the approach and support for parents of babies who may receive a positive screen, including false positives.

"The opportunity to identify severe combined immunodeficiency early in life has transformed outcomes for this otherwise fatal condition. Introducing this assay into newborn screening programs throughout Canada has the potential to save lives and prevent suffering of patients and families affected by this condition," the authors conclude.

The review was created by a pan-Canadian group of physicians from BC Children's Hospital, University of British Columbia, Vancouver, BC; University of Montreal and CHU Sainte-Justine, Montréal, Quebec; IWK Health Centre and Dalhousie University, Halifax, Nova Scotia; and The Hospital for Sick Children and the University of Toronto, Toronto, Ontario.

More information: Catherine M. Biggs et al. Newborn screening for severe combined immunodeficiency: a primer for clinicians, *Canadian Medical Association Journal* (2017). DOI: 10.1503/cmaj.170561

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