

The challenge of severe infections in infancy

A five-month-old Caucasian female infant born at term weighing 3.5 kg was admitted with respiratory distress. Starting at 2.5 months of age, she suffered from oral thrush and chronic diarrhea, followed by cough, wheezing and fatigue. She was below the third percentile for height and weight (58 cm and 4.2 kg, respectively). Her laboratory evaluation showed a total lymphocyte count of 900 with low lymphocyte subsets, neutropenia and eosinophilia; low immunoglobulins; and negative HIV polymerase chain reaction and sweat chloride tests. Her chest x-ray had

evidence of pneumonitis and an absence of thymus tissue. The respiratory syncytial virus (RSV) screening test was positive and purine/pyrimidine studies revealed adenosine deaminase (ADA) deficiency, confirming a diagnosis of severe combined immunodeficiency (SCID). She was treated with trimethoprim-sulfamethoxazole prophylaxis, intravenous immunoglobulins and enzyme replacement, and underwent bone marrow transplantation. Despite intensive care management, chronic RSV infection complicated her transplant and she died.

LEARNING POINTS

- Between April 2004 and August 2005, the SCID study of the Canadian Paediatric Surveillance Program has confirmed nine cases to date.
- SCID is a serious, life-threatening condition with high morbidity and mortality.
- Bacille Calmette-Guérin (BCG) vaccination is contraindicated for infants and children with immunodeficiency diseases because of severe complications, such as osteomyelitis and disseminated BCG infection.
- Great advances have been made in identifying a variety of molecular genetic defects causing SCID. The two most common forms are X-linked SCID (approximately 50% of cases) and ADA deficiency (approximately 15% to 25% of cases).
- Effective therapeutic interventions, including bone marrow and stem cell transplantations, are now available for patients with SCID.
- The challenge in infants with SCID and the most important prognostic factor rely on confirming the diagnosis and performing bone marrow transplantation *before* the appearance of overwhelming infections.
- For further information on SCID and details as to appropriate management, including bone marrow transplantation, visit the Immune Deficiency Foundation's Web site at <www.primaryimmune.org/>

The Canadian Paediatric Surveillance Program (CPSP) is a joint project of the Canadian Paediatric Society and Public Health Agency of Canada that undertakes the surveillance of rare diseases and conditions in children. For more information, visit our Web site at <www.cps.ca/cpsp> or <www.cps.ca/pcsp>.