

WINDOWS ON HADASSAH

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The transformation of Miley Hyland from a sick infant (above) to healthy toddler (left).

JERUSALEM GENETIC DIAGNOSIS EASES AUSTRALIAN FAMILY'S ANXIETY

Until recently, Sally and James Hyland of Queensland, Australia, had no idea why their daughter, Miley, suffered from liver failure and seizures. Their 18-month search for a diagnosis ended when Prof. Orly Elpeleg, Head of HMO's Department of Human Genetics and Metabolic Diseases, identified the problem. Miley has a mutation in the mitochondrial gene; the mutation causes liver failure in infants.

In September 2009, *The American Journal of Human Genetics* published the results

of Prof. Elpeleg's investigation into the phenomenon of babies, one to five months old, admitted to hospitals with severe liver failure. When she and her team analyzed and compared the DNA of the sick babies, they discovered an identical segment with more than 20 encoded genes. A mutation in the mitochondrial gene revealed a repetitive biochemical pattern.

The physician treating Miley at the Royal Children's Hospital in Brisbane consulted with a physician in Great Britain who referred them to Prof. Elpeleg. After receiving a sample of Miley's DNA, Prof. Elpeleg immediately identified the problem and informed the

parents that once children live beyond five months, they usually have no further liver problems. Miley had already passed that significant milestone.

"We are so grateful that we finally have the answers we were waiting so long for, thanks to you and Miley's doctors here in Australia," Sally Hyland wrote Prof. Elpeleg. "Fortunately the outcome wasn't as bad as we first expected and we are hoping that there will be no more dreadful liver failure/seizure activity."

Prof. Elpeleg's original research indicated that all the sick children she examined in Israel came from Jewish families of Yemenite origin. In the course of their research, she and her team also found a few sick children of Ashkenazi Jewish origin, as well as French children of Algerian origin and Arab children whose conditions were caused by other mutations in the same gene. Now it turns out a child in Australia carries yet another new mutation in the same gene.