

# 4.1

## CLINICAL APPLICATION

### OVERRIDING A BLOCK IN GLYCOLYSIS

**Michael P. was noticeably weak from his birth. He didn't move much, had poor muscle tone and difficulty breathing, and grew exhausted merely from the effort of feeding. At the age of two and a half months, he suffered his first seizure, staring and jerking his limbs for several frightening minutes. Despite medication, his seizures continued, occurring more frequently.**

The doctors were puzzled because the results of most of Michael's many medical tests were normal—with one notable

exception. His cerebrospinal fluid (the fluid that bathes the brain and spinal cord) was unusually high in glucose suggesting that Michael's cells were not adequately catabolizing glucose.

Hypothesizing that a profound lack of ATP directly caused the symptoms, medical researchers tried dietary intervention beyond the block in the boy's metabolic pathway, taking a detour to energy production. When Michael was seven and a half months old, he began a diet rich in certain fatty acids. Within four days, he appeared to be healthy for the

very first time! The diet had resumed cellular respiration at the point of acetyl coenzyme A formation by supplying an alternative to glucose. Other children with similar symptoms have since enjoyed spectacular recoveries similar to Michael's thanks to the dietary intervention, but doctors do not yet know the long-term effects of the therapy. This medical success story, however, illustrates the importance of the energy pathways—and how valuable our understanding of them can be. ■