This Week's Citation Classic
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The report describes the first patient known to have hereditary galactokinase deficiency, his equally affected sisters, and his family. It establishes that the disorder causes juvenile cataracts and galactose-galactitol-glucose diabetes. [The SCI® indicates that this paper has been cited in over 145 publications.]

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In 1956 in Berne, Carl Cori and Herman Kalckar lectured on disorders of carbohydrate metabolism. I was a young intern asked to present excerpts of my doctoral thesis. British colleagues reported the first cases of galactosemia, which we had never seen at the Children's Hospital in Zurich. Before long, I diagnosed our first case, began to read old records, and came upon those of a nine-year-old blind gypsy boy seen in 1932. The charts had been carefully handwritten by Guido Fanconi, then a young chairman of the department he was still heading. The boy, a presumed diabetic, had galactose diabetes, a unique anomaly Fanconi identified while studying the effects of a fruit carbohydrate metabolism. I was a young investigator. It also taught me that human mutations can be predicted. Indeed, the first mutations can be predicted. Indeed, the first mutation for the last three. It shed far more light on the pathogenesis of galactosemia than had two years of my previous work on dozens of galactose-poisoned rats. And of course, it had a mildly exotic flavor as a "gypsy disease." To date, of the 21 European children diagnosed in my lab, 16 are either gypsy, Bulgarian, Yugoslavian, or all three.

Although I have not received any awards for this work, discovery of galactokinase deficiency gave me credit as a researcher and helped my academic career. It also taught me that human mutations can be predicted. Indeed, the first mutant for the last three steps in the Leloir pathway was soon to be discovered.