

Health



## Clinician Story: Pediatric Resident Diagnoses Very Rare Case of Neonatal Diabetes Mellitus

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### THE CLINICAL CASE

The parents of a one-month old boy brought their child to the outpatient clinic at Anjo Kosei Hospital, where I was just starting my senior residency in the Department of Pediatrics.

The infant had been vomiting since the previous day and his breathing was labored. The boy weighed just over 7 pounds. He had lost approximately half a pound over the last seven days.

The child was very sick due to severe dehydration, and he had an uncompensated metabolic acidosis (pH 6.980, BE-25.5). Additional laboratory testing revealed hyperglycemia (glucose = 706 mg/dL) and ketosis (total ketone bodies = 13750  $\mu\text{mol/L}$ ), which suggested diabetic ketoacidosis. He could have died within several hours if left untreated.

### DIAGNOSIS AND TREATMENT

The staff professionals I consulted had never seen such a young infant with this clinical presentation. If the child had been older, diabetic ketoacidosis associated with type 1 diabetes mellitus may have been suspected. However, type 1 diabetes mellitus is very rare in neonates and infants.

I wondered if the infant had a congenital metabolic disease or a severe infection. I accessed UpToDate on a computer at our clinic. I searched for a possible diagnosis and appropriate treatment.

UpToDate helped me reach the correct diagnosis at first consultation — neonatal diabetes mellitus — an exceedingly rare disease with an incidence rate of 1 in 500,000. This enabled us to provide the appropriate treatment and contact an external specialist without wasting any time. I also learned about the pathogenesis of neonatal diabetes from UpToDate and the appropriate work-up, and found that the child had a *KCNJ11* gene mutation.

I am happy to report that the child is thriving today. His blood glucose is stable on oral medication and he is doing fine with regular hospital visits. Each year, his parents send a seasonal greeting card with a photo, and I enjoy seeing his growth.

### TRUST IN UPTODATE

Since I had experience using UpToDate, I knew that I would be able to review an exhaustive list of differential diagnoses and diseases. I could proceed with appropriate treatment without hesitation because I was fairly confident that, based on the child's symptoms, it was unlikely to be a disease other than neonatal diabetes mellitus.

Using the information I found on UpToDate, I could also provide the parents with an explanation of the child's condition. This helped to reassure them that we were giving the right treatment.

Without access to UpToDate, it would have taken longer for me to reach the right

diagnosis. Insulin treatment could have been delayed and the patient might have suffered severe consequences, even death.

In general, a pediatric department is rather unique because patients show up with all types of complaints. We often see patients with underlying diseases or rare syndromes. Even now, as a teaching doctor, I always consult UpToDate to review different diseases and differential diagnoses.

In this era of information overload, high-quality information curated from the viewpoint of clinicians is really valuable. For me, UpToDate has been a reliable companion from my days in training as a medical student and a resident, to the present, where I am a specialist and a teaching doctor. I would recommend UpToDate to a colleague with confidence.

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