Transient neonatal diabetes mellitus due to paternal uniparental disomy of chromosome 6.

Fudvoye J. (1), Farhat K. (2) and Nicolescu R. (3)

(1): Pediatric department, CHU Liège
(2): CHU Liège
(3): Pediatric Endocrinology and Diabetology Department, Centre Hospitalier de la Citadelle, Liège

We present a case of neonatal diabetes diagnosed in the first week of life.

A 4-day-old newborn boy was referred to our Neonatal Intensive Care Unit for high blood sugar levels on repetitive samples. His gestational age was 37 weeks and he presented a severe intra-uterine growth restriction (IUGR) with birth weight 1980 g, (< Percentile 3), length 43 cm, (< Percentile 3) and head circumference 30 cm, (< Percentile 3 on WHO chart). Except the severe IUGR and a mild macroglossia, clinical examination was normal.

Routine blood sugar monitoring for IUGR babies reveals hyperglycemia (180 mg/dl) and glycosuria, without ketonuria. Insulin/C-peptide blood levels measured at the time of hyperglycemia were particularly low (insulin 1.4-1.6 mUI/l, C-peptide 0.14-0.16 nmol/l, blood sugar 229 mg/dl). The abdominal ultrasound did not visualize the pancreas (abdominal distension).

The diagnosis of neonatal diabetes mellitus was made and treatment by a short-acting insulin analogue was started (with initial intravenous delivery during the first 10 days and after then switch to a continuous subcutaneous infusion).

The first genetic tests looking for genes frequently associated with permanent neonatal diabetes mellitus (INS, KCNJ11 and ABCC8) found no mutation. The clinical impression of evolution to a transient diabetes (decreasing needs of insulin with definitive stop at the age of 8 weeks) was soon confirmed by genetic testing showing a complete loss of methylation on chromosome 6q24 (paternal uniparental isodisomy).

At the age of 3 months, his clinical (anthropometric, neurodevelopmental) and biological (blood sugar levels) parameters are strictly normal.