**Permanent Neonatal Diabetes Due to NKX2-2 Mutation: First report from the UAE**

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Neonatal diabetes mellitus (NDM) occurs in the first six months of life and is classified based on clinical outcomes. Permanent NDM (PNDM) is generally considered if insulin requirement persists up to 18 months of age. The incidence of PNDM and transient NDM in the Emirate of Abu Dhabi, UAE has been reported to be 1:31,900 and 1:350,903, respectively. Here we present the case of a patient with PNDM who was found homozygous for NKX2-2 nonsense variant p.(Arg129Ter) which, to the best of our knowledge, is the first reported case in the UAE.

KA is an Emirati female delivered via caesarean section with severe intrauterine growth retardation (1.2kg at 38 weeks). Her mother was diagnosed as gestational diabetes requiring insulin therapy. On postnatal day 7, she was a poor feeder, lethargic, and dehydrated. Her blood glucose was high (>30 mmol/L) and she was commenced on insulin therapy. At 3 years of age, she was extremely obese (weight 17 kg, >97th percentile); brain CT-scan showed normal brain parenchyma and ventricles; EEG showed abnormal wake state with generalized slow background activity. Of note were previous medical history significant of seizures, developmental delay and reduced visual acuity. Investigations revealed negative GAD (<5 IU/mL) and IA2 (<10 IU/mL) antibodies. KA, now 19-years of age, obese (87.4 kg) with poorly-controlled glycaemia (HbA1C ~8%), is wheelchair-bound due to paraparesis. Following flash glucose monitoring, insulin regimen was adjusted (insulin aspart 12-16 units; insulin degludec 24-26 units daily); HbA1C reduced to 7.4%, with less glycaemic variability. Medical management comprises atorvastatin 40mg OD, lisinopril 10 mg OD, metformin 1000mg BID, and liraglutide. Genetic testing has been recommended for her parents to confirm their carrier status.

The reported case showed characteristic attributes of PNDM due to NKX2-2 mutation with severe neurologic features. The latter have not been previously reported on other cases. All cases of diabetes diagnosed before 6 months of age, particularly those with features described here should be considered of this potential diagnosis.