Neonatal Diabetes – The Great Masquerader

Experiences from one hospital

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Introduction

Neonatal diabetes (NDM) is diagnosed within the first six months of life.

The incidence is 1 in 400,000 live births.

NDM can often be confused with sepsis as there is considerable overlap of symptoms in this age group as illustrated in the case series that we present below.

In both infants the diagnosis could have been missed due to the non specific nature of the presenting symptoms.

We recommend an initial check of blood glucose concentrations in all sick infants who present to Accident and Emergency.

Case 1

A 7 week old, born to nonconsanguineous parents presented with a temperature of 38.6C and a one day history of poor feeding

Clinical examination was unremarkable. The working diagnosis was possible sepsis.

A blood gas that was done for monitoring of acid base status showed a glucose concentration of 39mmol/l with a normal ph.

She was commenced on an insulin infusion then commenced on Continuous Subcutaneous Insulin Infusion (CSII)

Genetic analysis showed a KCNJ11 mutation. Insulin was stopped and Glibenclamide was started. She responded well and currently has good glycaemic control.

Case 2

A second baby, born to consanguineous parents (birth weight of 2.7kg) presented at 24 days of age with a one day history of vomiting and poor feeding.

He was mildly tachypnoeic on arrival and a presumptive diagnosis of sepsis was made.

Interestingly, again an incidental blood gas revealed the infant to be in ketoacidosis with a blood glucose of 43mmol/L.

Insulin infusion was commenced and the baby was subsequently managed on CSII

Genetic analysis revealed a recessive non coding *INS* mutation. He went into remission in two weeks and is currently off insulin.

Conclusions

The cases described above illustrate the importance of blood glucose monitoring in sick infants presenting to emergency care settings.

In both infants the diagnosis could have been missed due to the non specific clinical presentation.

We recommend an initial check of blood glucose concentrations in all sick infants who present to A&E.

Identifying the underlying genetic mutations guides treatment and prognosis. The phenotype of the diabetes in both infants correlated with the respective mutations.

We concur with the policy of central genetic testing in these patients and suggest that CSII is the therapeutic intervention of choice.

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Diabetes 2

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