

# SINGLE CENTRE EXPERIENCE OF NEONATAL DIABETES

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## INTRODUCTION

Neonatal diabetes is a relatively uncommon disorder that can often present to the clinician with myriad manifestations and variable outcomes and can often be misdiagnosed as Type 1 diabetes. Hence, in this study we set out to study the correlation of disease with clinical features and genetics, suitability of current treatment regimens and treatment outcomes of 6 children presenting to a single centre in South India.

## Methods

Neonatal diabetes was defined as "A Diagnosis of Diabetes prior to 9 months of age. "Ages at recruitment ranged from 5 days to 18 years. Detailed history and investigations were obtained and included HbA1C, blood glucose, liver and renal function tests and autoimmune markers. Genetic samples were sent to the Genetics Lab of University of Exeter Medical School.

### CASE 1

7 day old male infant, was referred to the Neonatal unit with intractable seizures since Day 4 of life.

Evaluated and found to have an RBS of 33 mmol/l (594 mg/dl) and urine ketones 2+.

MRI brain revealed a large cerebral hemorrhage/

hemorrhagic infarct in the right fronto-temporo-parietal region with mass effect and shift of midline structures to the right.

He was started on insulin infusion and genetic samples were sent.

The neonate was continued on ventilation, multiple anti-epileptics and supportive care. However, the seizures proved to be intractable.

In view of the poor general condition of the neonate and poor prognosis, his parents consented to taking him off life support.

Genetic testing revealed a novel novel KCNJ11 missense variant, p.R29H inherited along the maternal line, but causal association with neonatal diabetes could not be proven.

**To the best of our knowledge, this is only the second case of neonatal diabetes presenting with a cerebral infarct**

### CASE 2

19 year old who presented for management of diabetes.

Was diagnosed at 8 months of age with an initial Blood Glucose of 20 mmol/l (360 mg/dl) and diabetic ketoacidosis.

Has been on insulin since diagnosis and is now on 0.8 units/kg/day.

Genetic evaluation revealed an INS missense mutation, located at Exon3, p.R89C.

She is presently being managed with insulin, as earlier.

### CASE 4

14 month old male diagnosed at 4 months of age when he presented with shortness of breath and an RBS of 29 mmol/l (524 mg/dl).

He was being managed on insulin at presentation.

Genetic evaluation revealed heterozygosity for an ABCC8 nonsense mutation, p.W232, and an ABCC8 missense mutation, p.P254S. The p.W232\* mutation is predicted to be pathogenic and testing has shown that the child has inherited this mutation from his unaffected mother and transition to sulphonylurea could be successful.

Father was not available for genetic analysis.

Transition was partially successful, but he still needs a small dose of basal insulin at bedtime

### CASE 6

A 14 year old male child who was diagnosed at 9 months of age with bronchopneumonia and diabetic ketoacidosis.

Was being managed on insulin.

Parents are non-consanguineous and there is no family history of diabetes

Genetic evaluation was negative for all known mutations of neonatal diabetes.

He is being managed on insulin.

DNA samples have been preserved for future testing.

## CONCLUSION

Neonatal diabetes can often present with diabetic ketoacidosis and can be mistaken for Type 1 diabetes. With proper clinical vigilance and timely genetic analysis, basic research findings can be translated into accurate treatment decisions and good clinical outcomes in neonatal diabetes and especially, the outcomes of transition onto sulphonylurea can be improved.

## REFERENCES

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