TYPE 5 MONOGENIC DIABETES: REPORT OF 7 CASES.

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INTRODUCTION:

Type 5 monogenic diabetes is an autosomal dominant disease due to a mutation in *HNF1beta* gene. This gene is expressed predominantly in kidney and pancreas, thus the clinical manifestations are characterised by renal abnormalities and diabetes.

OBJECTIVES:

To review the clinical characteristics of patients who were diagnosed with type 5 monogenic diabetes in the Pediatric Endocrinology Unit of a tertiary referral hospital.

METHODS:

The medical history of 7 children who were diagnosed with type 5 monogenic diabetes with genetic confirmation were reviewed.

RESULTS:

Patient information is described in the table attached.

	Sex	Prenatal history	Familial diabetes history	Symptomatology (age in years)	Age at genetic diagnosis	Genetic	HbA1c at genetic diagnosis (% 4.7-6.4)	Age at Diabetes diagnosis (years)	Renal	Other pathologies	Current Diabetes treatment
1	M	Intrauterine growth restriction Renal cysts	-Maternal grandfather with Type 2 Diabetes	Renal anomaly (1)	13	Heterozygous total gene deletion	6.7	13	Bilateral renal subcortical cysts.	None reported	Insulin
2	M	Intrauterine growth restriction	-Maternal grandmother with Type 2 Diabetes	Diabetic ketoacidosis (15)	15	Heterozygous total gene deletion	14	15	Bilateral renal dysplasia with multiple cysts.	Hypertransaminasemia	Insulin
3	M	Intrauterine growth restriction	-Father with hypercholesterolemia -Maternal grandfather with Type 2 Diabetes	Hypertransaminasemia (5)	18	Heterozygous total gene deletion	5.8	20	Bilateral renal dysplasia. Left kidney with multiple medular cysts.	Hypertriglyceridemia, Growth retardation	Insulin
4	F	Intrauterine growth restriction	-Maternal grandfather with Type 2 Diabetes	Hyperglycemia Proteinuria (13)	13	Exon 2: c 513> C p. Trip171cys Heterozygous mutation	6.5	13	At diagnosis: normal kidney ultrasound 17 years old: cortical and subcortical cysts. Currently: stage III RCF	Hyperuricemia	None
5	F	None reported	-Maternal grandfather with Type 2 Diabetes	Renal anomaly (1)	12	Heterozygous total gene deletion	6.2	17	At diagnosis: renal glomerulocystic bilateral dysplasia. Evolution to renal failure. Currently 2on kidney transplant	Exocrine pancreatic insufficiency	Insulin
6	M	Intrauterine growth restriction	-Maternal grandfather with Type 2 Diabetes	Hypertransaminasemia (8)	16	Heterozygous total gene deletion	5.1	No diabetes	Little multiple cortical bilateral cysts.	Hypomagnesemia	None
7	M	Intrauterine growth restriction Left renal dysplasia	None reported	Renal anomaly (1)	14	Heterozygous total gene deletion	___	No diabetes	At diagnosis: Righ monorenal Currently: Stage II RCF	Growth retardation, Hypertransaminasemia Hypomagnesemia	None

CONCLUSION:

Phenotypic variability at the onset of Type 5 monogenic diabetes implies a diagnostic challenge.

The study of *HNF1beta* gene should be considered in any patient with hyperglycemia, negative antibodies for diabetes, family history of type 2 diabetes and renal abnormalities.

While progressive dysfunction of beta cells is observed, not all the patients require insulin treatment at the beginning of the disease.

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