Have you tried google?

Grant F. King

R.C.A.D. Patient Day

U.C.L., Sept. 2012

A NICE DAY OUT…

- “Bad kidneys”
- Drink plenty!
- The curse of cotton wool!
- Maternal guilt

97 NOT OUT!

- *The Worried Unwell*

- *"An emergency admission was recorded at 17 years with a 2-week history of osmotic symptoms, laboratory glucose of 97 mmol/L with a normal bicarbonate level and no evidence of ketoacidosis."*

"The admitting physician was perplexed by 'the lack of ketoacidosis in someone of his age' and wondered as to why he presented in a 'hyperglycaemic non-ketotic state' rather than with diabetic ketoacidosis."

- *Insulin- Just Take It!*

The demon drink(s)

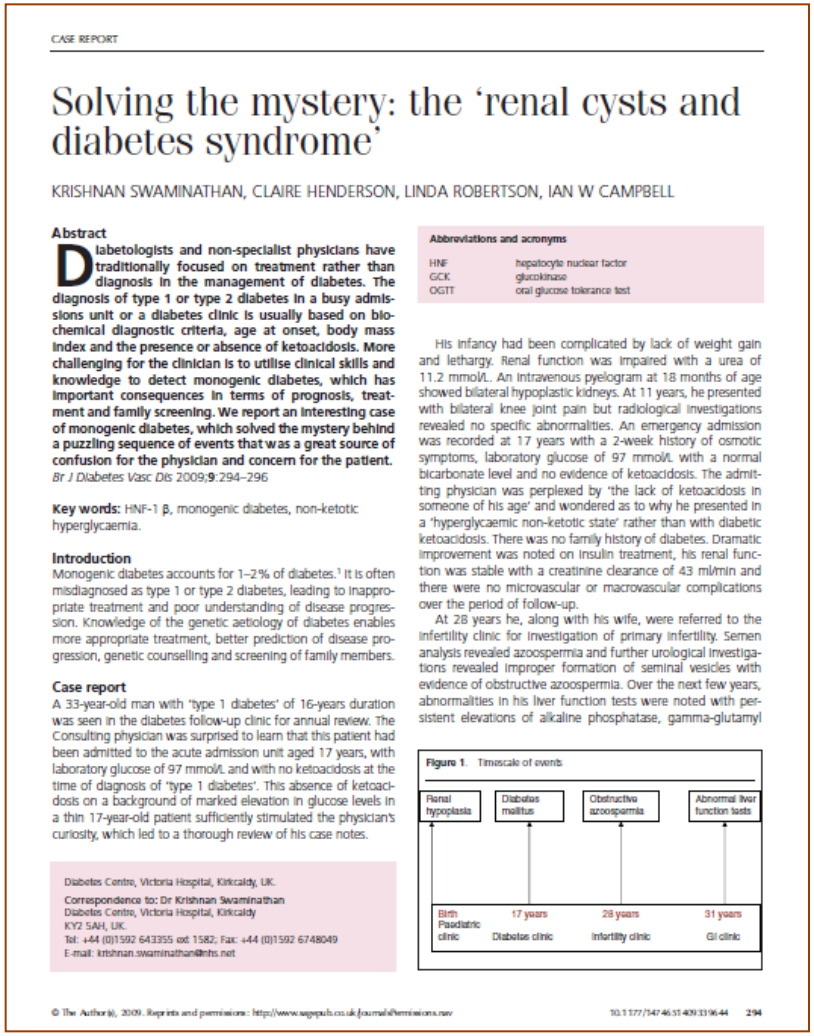
- Moo-re problems…

- Boozey Biopsy

Finding & Facing Fertility issues…

Have you tried google?

- Solving the Mystery!



CASE REPORT

Solving the mystery: the 'renal cysts and diabetes syndrome'

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Abstract

Diabetologists and non-specialist physicians have traditionally focused on treatment rather than diagnosis in the management of diabetes. The diagnosis of type 1 or type 2 diabetes in a busy admissions unit or a diabetes clinic is usually based on biochemical diagnostic criteria, age at onset, body mass index and the presence or absence of ketoacidosis. More challenging for the clinician is to utilise clinical skills and knowledge to detect monogenic diabetes, which has important consequences in terms of prognosis, treatment and family screening. We report an interesting case of monogenic diabetes, which solved the mystery behind a puzzling sequence of events that was a great source of confusion for the physician and concern for the patient.

Br J Diabetes Vasc Dis 2009;**9**:294–296

Key words: HNF-1 β, monogenic diabetes, non-ketotic hyperglycaemia.

Introduction

Monogenic diabetes accounts for 1–2% of diabetes.[1] It is often misdiagnosed as type 1 or type 2 diabetes, leading to inappropriate treatment and poor understanding of disease progression. Knowledge of the genetic aetiology of diabetes enables more appropriate treatment, better prediction of disease progression, genetic counselling and screening of family members.

Case report

A 33-year-old man with 'type 1 diabetes' of 16-years duration was seen in the diabetes follow-up clinic for annual review. The Consulting physician was surprised to learn that this patient had been admitted to the acute admission unit aged 17 years, with laboratory glucose of 97 mmol/L and with no ketoacidosis at the time of diagnosis of 'type 1 diabetes'. This absence of ketoacidosis on a background of marked elevation in glucose levels in a thin 17-year-old patient sufficiently stimulated the physician's curiosity, which led to a thorough review of his case notes.

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Abbreviations and acronyms

HNF	hepatocyte nuclear factor
GCK	glucokinase
OGTT	oral glucose tolerance test

His infancy had been complicated by lack of weight gain and lethargy. Renal function was impaired with a urea of 11.2 mmol/L. An intravenous pyelogram at 18 months of age showed bilateral hypoplastic kidneys. At 11 years, he presented with bilateral knee joint pain but radiological investigations revealed no specific abnormalities. An emergency admission was recorded at 17 years with a 2-week history of osmotic symptoms, laboratory glucose of 97 mmol/L with a normal bicarbonate level and no evidence of ketoacidosis. The admitting physician was perplexed by 'the lack of ketoacidosis in someone of his age' and wondered as to why he presented in a 'hyperglycaemic non-ketotic state' rather than with diabetic ketoacidosis. There was no family history of diabetes. Dramatic improvement was noted on insulin treatment, his renal function was stable with a creatinine clearance of 43 ml/min and there were no microvascular or macrovascular complications over the period of follow-up.

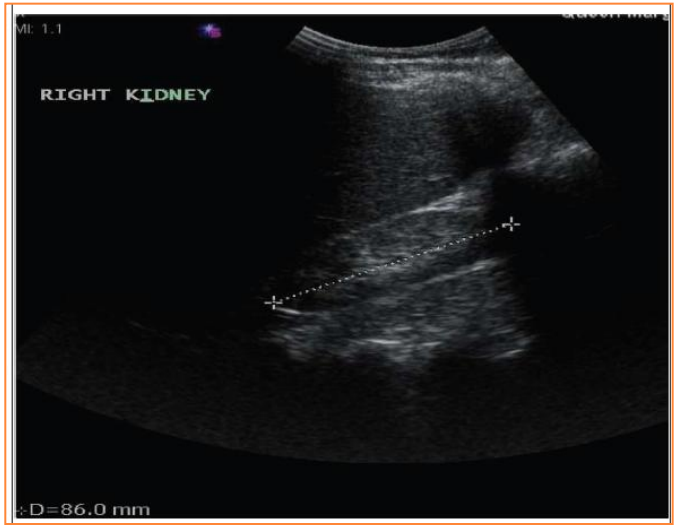
At 28 years he, along with his wife, were referred to the infertility clinic for investigation of primary infertility. Semen analysis revealed azoospermia and further urological investigations revealed improper formation of seminal vesicles with evidence of obstructive azoospermia. Over the next few years, abnormalities in his liver function tests were noted with persistent elevations of alkaline phosphatase, gamma-glutamyl

Figure 1. Timescale of events

10.1177/1474651409339644

Stent and stones

- Ouch!
- Surgery
- Lemonade therapy?



Spontaneity

Bits and bobs

- Thyroid
- Diverticular disease
- Knees

- Tired!
- Work (perspiration and inspiration)
- Insurance
- Invisibility

What helped?

- Relationships
- Reality
- Rest
- RD clinic

Thanks for listening!