# Updates on Two Interesting Cases: Case 2

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## 24y/o F with polyuria, blurred vision, and fatigue

 24 y/o Filipino woman sent by PCP with c/o polyuria, blurred vision, and fatigue

Last PCP visit, fasting BG 171, HbA1c 7.5%

Had not started medication

#### **Patient History**

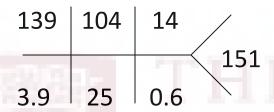
- PMHx
  - Seasonal allergies
  - GERD
- Meds
  - Loratadine 10mg PO daily
  - Nasonex two sprays each nostril daily
  - Prevacid 30mg PO BID
- Allergies
  - Lactose
  - NKDA

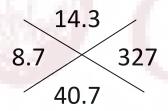
- FamHx
  - MGM: DM2 in 80's
  - F: prediabetes, asthma, seasonal allergies
  - M: Sjogren's disease,
     Rheumatoid arthritis, ASD
  - S: gestational diabetes
  - B: healthy
- SocHx
  - Single, no children
  - Graduate student in pediatric clinical neuropsychology
  - Denies tobacco, IVDU
  - Occasional EtOH

#### Physical Exam

- VS: BP 102/74 mm Hg, HR 68 bpm, Ht 61 in, Wt 57 kg, BMI 24, office fasting BG 146
- HEENT: oropharynx without crowding, thyroid normal
- Pulm: CTA bilaterally
- CV: RRR, nl S1, S2, no M/R, pedal pulses palpable, no edema
- Abd: soft, non-tender, non-distended, NABS
- MS: normal muscle strength 5/5 throughout
- Skin: no acanthosis nigricans, no skin tags
- Neuro: Sensation intact in b/l feet

#### **Laboratory Data**





Hgb A1c: 7.5%

GAD65 Ab: 0.00 nmol/L

IA2: negative

Insulin Ab: <3% bound

Glucose: 149 mg/dL

C-peptide: 0.7 pmol/mL

Microalbumin: 6.8 mcg/mg

TSH: 2.2 mcU/ml

T4: 7.9 mcg/dL

FTI: 9.2

#### Plan

Sample sent to test for MODY mutations

 Started pt on Januvia 100mg daily due to concern for beta cell dysfunction

Added Metformin 500mg BID to improve fasting blood glucose

#### **DNA** Testing

- MODY3 mutation found
  - exon 8 E508K mutation in HNF1A gene

### CHICAGO MEDICINE

	Genetic Defect	Result
MODY1	HNF-4α	Impairment of glucose- stimulated insulin secretion
MODY2	Glucokinase gene	Reduced activity of glucokinase, decreased insulin secretion
MODY3	HNF-1α	Weak transactivator of the insulin gene, decreased insulin secretion
MODY4	Insulin promotor factor-1	Abnl transcriptional regulation of beta cell development and function
MODY5	HNF-1β	Leads to early onset diabetes, kidney cysts
MODY6	NeuroD1	Regulatory switch for endocrine pancreatic development

#### **Treatment**

 No improvement in BG with Januvia and Metformin

- Patient started on Glyburide 5mg BID
  - Titrated to Glyburide 30mg BID, with improved BG control (HbA1c 6.8%)

#### Follow Up

- BG uncontrolled, Glyburide 40mg BID, started Lantus 8 units qhs
  - Titrated to Lantus 14units qhs
- A1c 7.4%
  - Started Januvia 100mg daily, decreased Lantus to 8 units qhs
- A1c 7.6 8.0%
  - Continued to increase insulin dosing (Lantus 28 units BID)
  - Multiple dietary changes made to avoid mealtime insulin
- Increasing patient frustration as A1c not improving, BG uncontrolled

#### Follow Up

- Felt Januvia was not helping, glyburide only medication keeping BG relatively stable
- Changed regimen to:
  - Glyburide 40mg BID
  - Lantus 28 units BID
  - Victoza 0.6mg daily -> titrate to 1.2mg daily
  - Stop Januvia

#### Follow Up

Patient emailed regarding hypoglycemia in the afternoon and evening

Decreased glyburide to 40mg qPM, 30mg qAM

 Now with improvement in blood glucose without hypoglycemia

Awaiting A1c at next visit