Case Study - Ian

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- Conflicts of Interest
  - None
    - Heather Rush

A conflict of interest exists when an individual is in a position to profit directly or indirectly through application of authority, influence, or knowledge in relation to the affairs of PENS. A conflict of interest also exists if a relative benefits or when the organization is adversely affected in any way.

Conflict of Interest Disclosure

- 3 y/o Ian

Kelly, mother of Ian, has given written permission for his picture and case to be presented to the PENS 2015 conference.

Objectives

- Describe the considerations without a gallbladder.
- Explain the teaching strategies to help the family understand diabetes management.
- Discuss take away points from this case study.

Diabetes complicated by past medical history

Birth history

- 31 w gestation fraternal twin
- Birth weight 1750 gm
- Respiratory distress at delivery requiring intubation
- Duodenal obstruction – repaired
- Cardio echo showed small PFO, moderate PDA, with moderate pulmonary hypertension
- Renal ultrasound – solitary left kidney
- Vertebral & rib anomalies

*Genetic consult

* Multiple congenital anomalies
* Genetics consult
* NORMAL MALE KARYOTYPE
* CGH – interstitial duplication of Xp 22.33 and 16p12.11 (? of clinical significance over time)

*Surgical procedures

* Laparotomy
* Appendectomy
* Ladd procedure
* Deudenoduodenostomy secondary to malrotation with duodenal atresia

*During hospitalization

* Klebsiella Pneumoniae sepsis
* Direct hyperbilirubinemia
* Right renal agenesis with left kidney mild hydronephrosis
* Spinal anomalies including 13 ribs (12 on left, 13 on right)
* Biliary atresia

*NICU stay

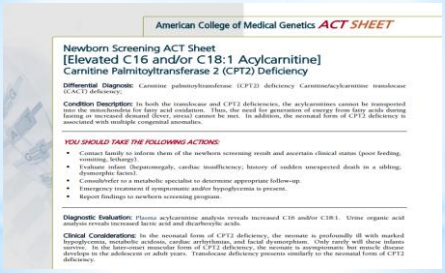
* Length of stay - 73 days
* Discharge weight 3340 grams; length 46.5 cm
* Cardio - PFO (f/u at 6 months of age)
* GI - appendectomy & duodenal resection; biliary atresia
  * Possible Alagille syndrome
* Renal - solitary left kidney (f/u at 4 months of age)

*Alagille syndrome

* Arteriohepatic dysplasia
* Autosomal-dominant
* Characterized by absence of intrahepatic bile ducts with progressive destruction of bile ducts, cholestasis, peripheral pulmonic stenosis, peculiar facies (broad forehead, deep-set eyes), cardiac lesions, vertebral arch defects, changes in renal tubules

*Endocrine issues

* Newborn screen -
  * Nl thyroid & biotinidase
  * Increased acylcarnitine level
* Free T4 & TSH at 25 days of life
  * 1.549 / 7.82
* Repeat newborn screen at 25 days of life
  * Decreased acylcarnitine with nl amino acids
* Received levocarnitine 15 mg/kg/day with TPN for 35 days
* Repeat acylcarnitine profile (7 days after treatment stopped) WNL with urine total & free carnitine WNL



American College of Medical Genetics ACT SHEET

Newborn Screening ACT Sheet

[Elevated C16 and/or C18:1 Acylcarnitine]

Carnitine Palmitoyltransferase 2 (CPT2) Deficiency

Differential Diagnosis: Carnitine palmitoyltransferase (CPT2) deficiency Carnitine/acylcarnitine translocase (CACT) deficiency;

Condition Description: In both the translocase and CPT2 deficiencies, the acylcarnitines cannot be transported into the mitochondria for fatty acid oxidation. Thus, the need for generation of energy from fatty acids during fasting or increased demand (fever, stress) cannot be met. In addition, the neonatal form of CPT2 deficiency is associated with multiple congenital anomalies.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- Evaluate infant (hepatomegaly, cardiac insufficiency, history of sudden unexpected death in a sibling; dysmorphic facies).
- Consult/refer to a metabolic specialist to determine appropriate follow-up.
- Emergency treatment if symptomatic and/or hypoglycemia is present.
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma acylcarnitine analysis reveals increased C16 and/or C18:1. Urine organic acid analysis reveals increased lactic acid and dicarboxylic acids.

Clinical Considerations: In the neonatal form of CPT2 deficiency, the neonate is profoundly ill with marked hypoglycemia, metabolic acidosis, cardiac arrhythmias, and facial dysmorphism. Only rarely will these infants survive. In the later-onset muscular form of CPT2 deficiency, the neonate is asymptomatic but muscle disease develops in the adolescent or adult years. Translocase deficiency presents similarly to the neonatal form of CPT2 deficiency.

*Interim history

* PDA closed on own with PFO closure by cardiology
* Absent gallbladder
* Cholestatic liver disease
* Abnormality of vertebrae - butterfly
* Absent right kidney - confirmed
* NL growth & development

Diabetes…

- 2 y 6 m
- 2-3 wk polyuria/polydipsia
- 1 day history of vomiting & decreased PO intake
- DKA – admitted to PICU

- Recovered within 24 hours
- Transitioned to Diabetes center floor for diabetes education
- Insulin started – Levemir BID & Humalog with meals

DKA admission

* A1C – 9.4% (H)
* C-peptide - 0.3 (L)
* GAD - 27.9 (H)
* Insulin antibodies – 11.6 (H)
* Celiac antibodies – neg
* Thyroid antibodies – neg
* TFTs – WNL

*Labs @ onset

* 10 days post hospitalization
* BG levels elevated
* Levemir 3 units AM, 2 units PM
* Humalog cartridge 1:70 with 1:200 > 150
* A1C – 10.1%
* Insulin doses changed to:
  * Lantus 5 units at supper
  * Humalog 1:60 for all meals (corrections remain the same)

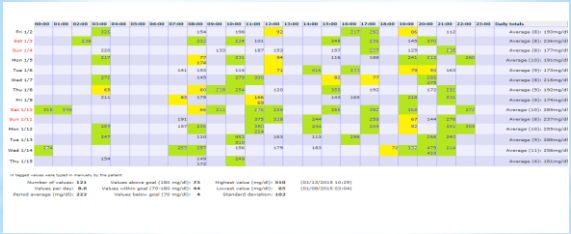
*Outpatient clinic

*Always elevated at 2 hr post breakfast

*Mom feeding eggs/bacon or oatmeal with milk

*Gastroperesis ???

*Key points to consider



*Occurs in 1 : 1000 people

*30% have additional malformations – malrotation of the gut & renal agenesis (just to name a few)

*Healthy, no treatment needed for absent gallbladder

*Absent Gallbladder

* Without the gallbladder, bile is still produced in the liver, but the release is slow & continuous into the intestines – therefore when eating higher fat content in meals, may not be adequate amounts of bile in the intestine to properly handle the normal absorption process.

* Low-fat
* High fiber
* KEY to REMEMBER = slow digestion

* Dietary considerations

* Insulin administration??
* Insulin pump??
* CGM??

* Thoughts???

*Questions??

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*References

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