Pancreatitis in a 28-Year-Old Woman
CASE PRESENTER: DISCUSSANTS:

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Disclosures:
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CASE PRESENTATION

28-year-old female

PMH/PSH:
Multiple episodes of pancreatitis since 3 mo. old.
Diagnosed with LPL deficiency by heparin assay.
Well controlled with diet during childhood.
In early twenties started to have recurrent bouts of pancreatitis.
15 pancreatitis hospitalizations over 4 year period, likely due to insurance and follow up issues.
Referred to our Lipid clinic by PCP.
CASE PRESENTATION

Social history:
Denies alcohol use, smoking. Uses marijuana occasionally.

Family history:
Father with elevated TG and chronic pancreatitis.
Brother with elevated TG and episodes of pancreatitis.
Mother with elevated TG.
Sister had first pancreatitis episode during pregnancy.
CASE PRESENTATION

Physical exam and vitals are unremarkable.
BMI 20.4

Labs:
T. Chol 114, TG 580, HDL 13 mg/dL, direct LDL 25
VLDL 76. TG ranges from 1000-2500.
CMP, TSH, UA NL, negative urine pregnancy test.
CT abdomen: pancreatitis with necrosis.
Medications: low fat diet
fenofibrate 160 mg/d, Ω-3FA 4000mg/d,
intermittently niacin.
FAMILY PEDIGREE

1st generation

TG1

TG3

c.644G>A(p.G215E)
c.708delA (p.G237fs*15)

2nd

TG5

c.644G>A(p.G215E)
c.708delA(p.G237fs*15)

TG6

TG4

c.708delA(p.G237fs*15)

3rd

PROBAND

c.644G>A(p.G215E)
c.708delA(p.G237fs*15)

TG2

c.644G>A(p.G215E)
## GENOTYPE-PHENOTYPE CORRELATION

<table>
<thead>
<tr>
<th>ID</th>
<th>MUTATION 1</th>
<th>MUTATION 2</th>
<th>RELATIONS</th>
<th>PANCREATITIS</th>
</tr>
</thead>
<tbody>
<tr>
<td>TG1</td>
<td>c.644G&gt;A (p. G215E)</td>
<td>NONE</td>
<td>MOTHER</td>
<td>NONE</td>
</tr>
<tr>
<td>TG2</td>
<td>c.644G&gt;A (p. G215E)</td>
<td>NONE</td>
<td>SON OF PROBAND</td>
<td>NONE</td>
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<tr>
<td>TG3</td>
<td>c.708delA (p. G237fs*15)</td>
<td>NONE</td>
<td>FATHER</td>
<td>2 EPISODES</td>
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<tr>
<td>TG4</td>
<td>c.708delA (p. G237fs*15)</td>
<td>NONE</td>
<td>BROTHER</td>
<td>SEVERAL EPISODES</td>
</tr>
<tr>
<td>TG5</td>
<td>c.644G&gt;A (p. G215E)</td>
<td>c.708delA (p. G237fs*15)</td>
<td>SISTER</td>
<td>ONE DURING PREGNANCY</td>
</tr>
<tr>
<td>TG6</td>
<td>NO</td>
<td>MUTATION</td>
<td>SISTER</td>
<td>NONE</td>
</tr>
<tr>
<td>PROBAND</td>
<td>c.644G&gt;A (p. G215E)</td>
<td>c.708delA (p. G237fs*15)</td>
<td></td>
<td>MULTIPLE EPISODES</td>
</tr>
</tbody>
</table>
CASE CONTINUED

With current medication regiment the patient was intermittently controlled.
She got pregnant at the age of 30 years old, was offered TPA nutrition or plasmapheresis. Patient was managed successfully with plasmapheresis through first pregnancy.

Currently, she is pregnant with twins and being managed with twice weekly plasmapheresis.
Sister had only one episode of pancreatitis during her pregnancy.
CONCLUSION

1. Phenotypic disease presentations of the same genotypes can vary significantly as it is seen in proband and her sister.

2. Some mutations are possibly more pathogenic than others: \textit{c.644G>A}(p.G215E) no episodes of pancreatitis in heterozygous individuals; \textit{c.708delA}(p.G237fs*15) even heterozygous family members had pancreatitis.

3. Management of affected individuals is challenging, especially during teenage years and during transition of care to adult lipid specialist.

4. Pregnancy can be successfully managed with plasmapheresis.