Clinical and Lab Findings at Diagnosis
Date of Summary: January 2013
Patients Enrolled: 361

PH Type

There are at least 3 forms of primary hyperoxaluria.

- Type I is caused by mutations of the AGXT gene. These mutations result in a deficiency of the enzyme alanine glyoxylate transferase (AGT) which is found only in the liver.

- Type II is caused by mutations of the GRHPR gene. These mutations result in a deficiency of the enzyme glyoxylate reductase/hydroxypyruvate reductase (GR/HPR) found in the liver and other tissues.

- Type III is caused by mutations of the HOGA1 gene (formerly DHDPSL) found in the liver.

<table>
<thead>
<tr>
<th>Type</th>
<th>Number</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>PH Type 1</td>
<td>265</td>
<td>73</td>
</tr>
<tr>
<td>PH Type 2</td>
<td>35</td>
<td>10</td>
</tr>
<tr>
<td>PH Type 3</td>
<td>31</td>
<td>9</td>
</tr>
<tr>
<td>Non PH1/PH2/PH3</td>
<td>24</td>
<td>7</td>
</tr>
<tr>
<td>Unknown</td>
<td>6</td>
<td>2</td>
</tr>
</tbody>
</table>

Of the 361 patients in the registry as of January 2013:

- 265 patients (73%) have Primary Hyperoxaluria Type I
- 35 patients (10%) have Primary Hyperoxaluria Type II
- 31 patients (9%) have Primary Hyperoxaluria Type III
- 24 patients (7%) do not have known mutations for Primary Hyperoxaluria I, II or III
Clinical and Lab Findings at Diagnosis

Date of Summary: January 2013
Patients Enrolled: 361

Country

PH Patients in Registry

PH patients enrolled in Registry from these countries
<table>
<thead>
<tr>
<th>Decade</th>
<th>Number</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>1960's</td>
<td>10</td>
<td>3</td>
</tr>
<tr>
<td>1970's</td>
<td>19</td>
<td>5</td>
</tr>
<tr>
<td>1980's</td>
<td>26</td>
<td>7</td>
</tr>
<tr>
<td>1990's</td>
<td>53</td>
<td>15</td>
</tr>
<tr>
<td>2000's</td>
<td>155</td>
<td>43</td>
</tr>
<tr>
<td>2010's</td>
<td>98</td>
<td>27</td>
</tr>
</tbody>
</table>

Of the 361 patients in the registry as of January 2013:

- 10 patients (3%) were diagnosed in the 1960’s
- 19 patients (5%) were diagnosed in the 1970’s
- 26 patients (7%) were diagnosed in the 1980’s
- 53 patients (15%) were diagnosed in the 1990’s
- 155 patients (43%) have been diagnosed since the year 2000
- 98 patients (27%) have been diagnosed since the year 2010
Symptoms and Findings | Percent at Diagnosis
--- | ---
Failure to Thrive | 8%
Nephrocalcinosis | 38%
Urolithiasis | 76%
Hematuria | 44%
Abdominal Pain | 58%
Asymptomatic | 11%

Of the 361 patients in the registry as of January 2013:

- 8% of the patients had failure to thrive (low height and weight) at diagnosis
- 38% of the patients had a history of nephrocalcinosis (when the kidneys are extensively filled with calcium oxalate crystals that can often cause loss of the kidneys.)
- 76% of the patients had a history of urolithiasis (kidney stones)
- 44% of the patients had a history of hematuria (blood in the urine)
- 58% of the patients had a history of abdominal pain
- 11% of the patients were asymptomatic (had no symptoms at all) at diagnosis

Some patients may have more than one symptom at diagnosis
Of the 361 patients in the registry as of January 2013:

- Approximately 150 patients had their first symptom between the ages of 0-4
- Approximately 50 patients had their first symptom between the ages of 5-9
- Approximately 60 patients had their first symptom between the ages of 10-19
  - Most patients have symptoms before the age of 25
Of the 361 patients in the registry as of January 2013:

- 95 patients were diagnosed between the ages of 0-4
- 65 patients were diagnosed between the ages of 5-9
- The chart shows that a large percentage of diagnoses are made before a patient’s 25th birthday
- Many patients are diagnosed before age 10
Of the 361 patients in the registry as of January 2013:

- The majority of patients have functioning kidneys at the time of diagnosis.