Clinical and Lab Findings at Diagnosis

Date of Summary: January 2013

Patients Enrolled: 361

PH Type

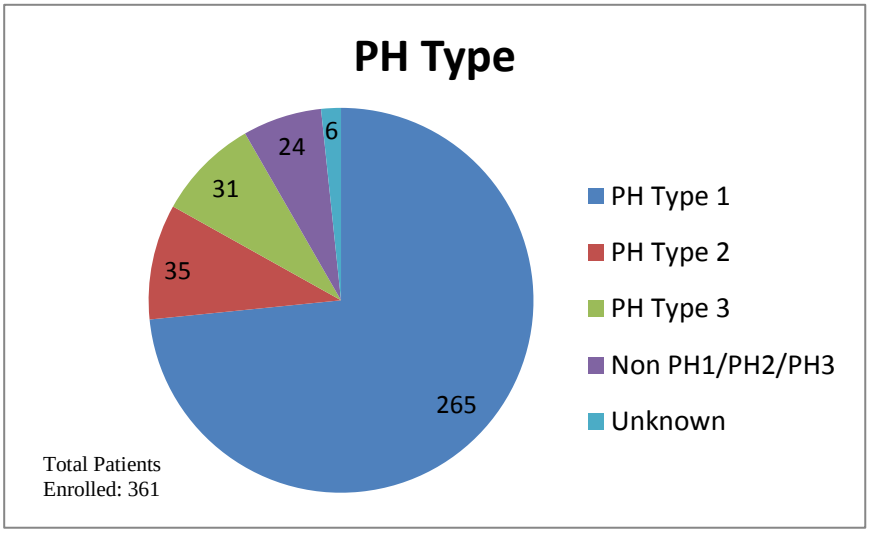
There are at least 3 forms of primary hyperoxaluria.

- **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.**

Type	Number	Percent
PH Type 1	265	73
PH Type 2	35	10
PH Type 3	31	9
Non PH1/PH2/PH3	24	7
Unknown	6	2

<u>Of the 361 patients in the registry as of January 2013:</u>

- **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**



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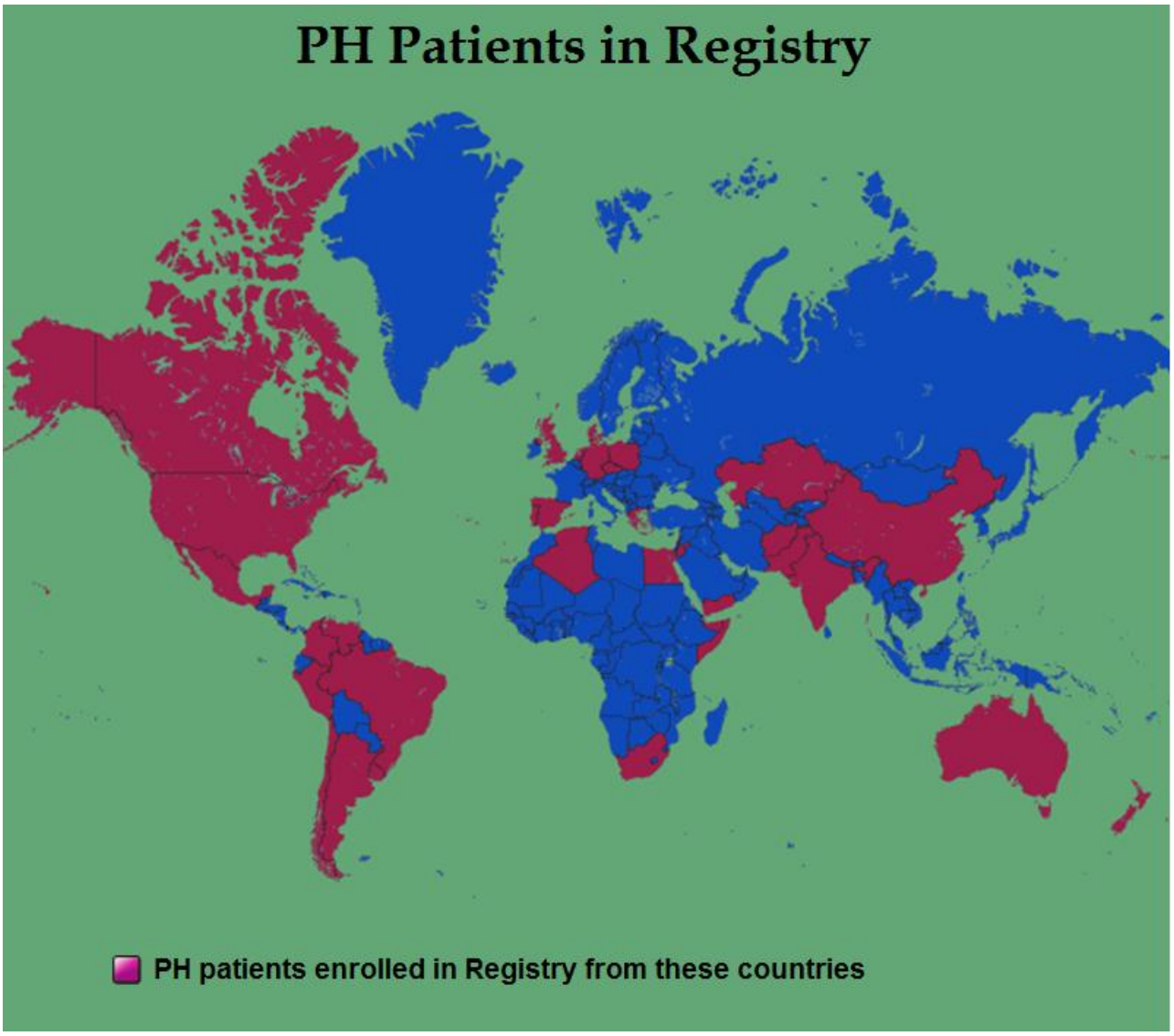
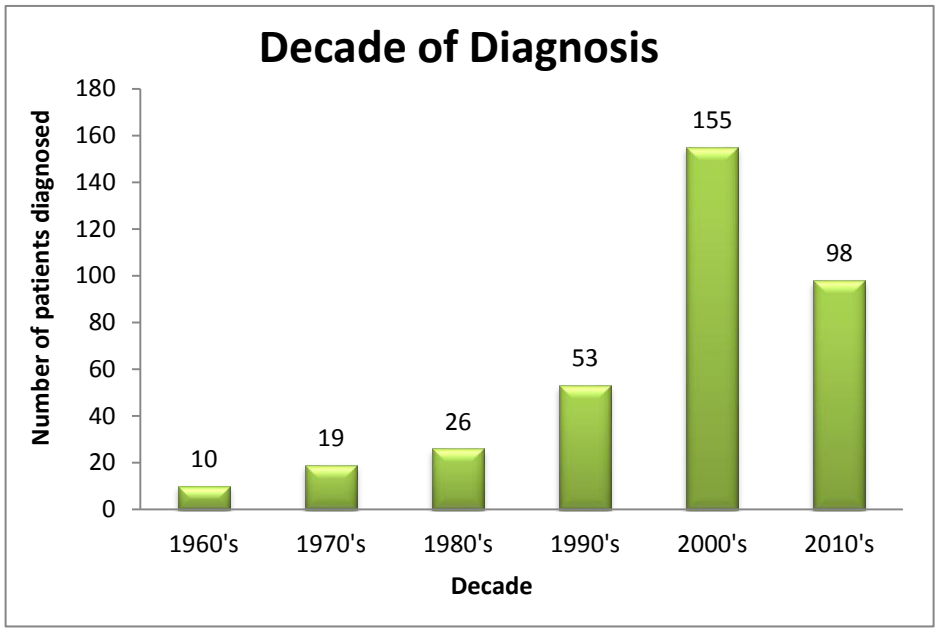


Decade	Number	Percent
1960's	10	3
1970's	19	5
1980's	26	7
1990's	53	15
2000's	155	43
2010's	98	27

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

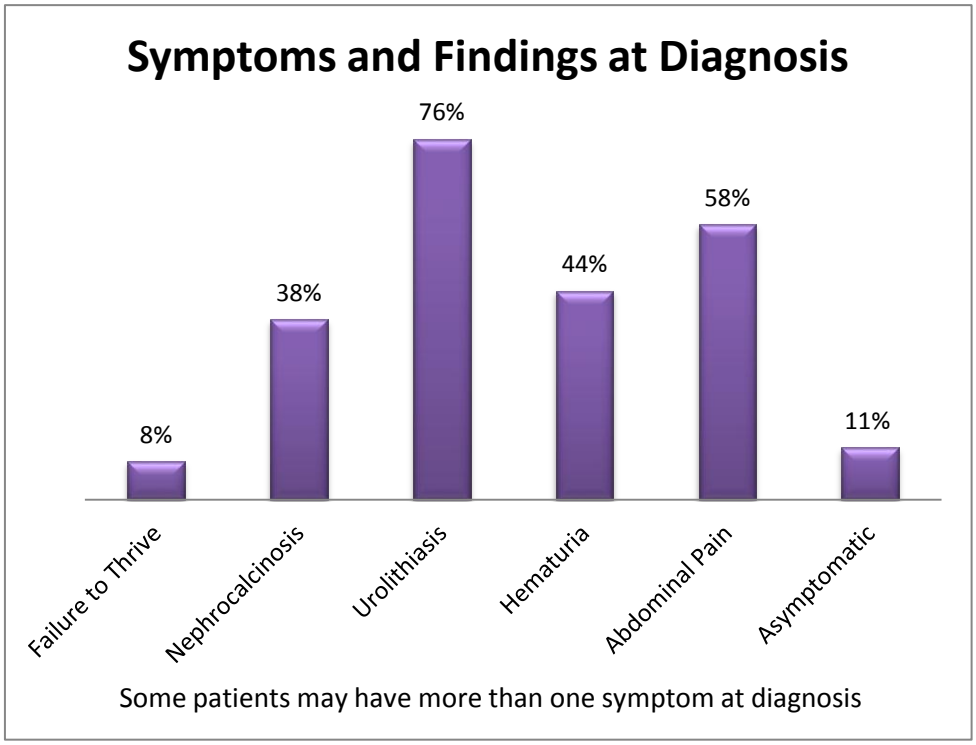
- **10 patients (3%) were diagnosed in the 1960's**
- **19 patients (5%) were diagnosed in the 1970's**
- **26 patients (7%) were diagnosed in the 1980's**
- **53 patients (15%) were diagnosed in the 1990's**
- **155 patients (43%) have been diagnosed since the year 2000**
- **98 patients (27%) have 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

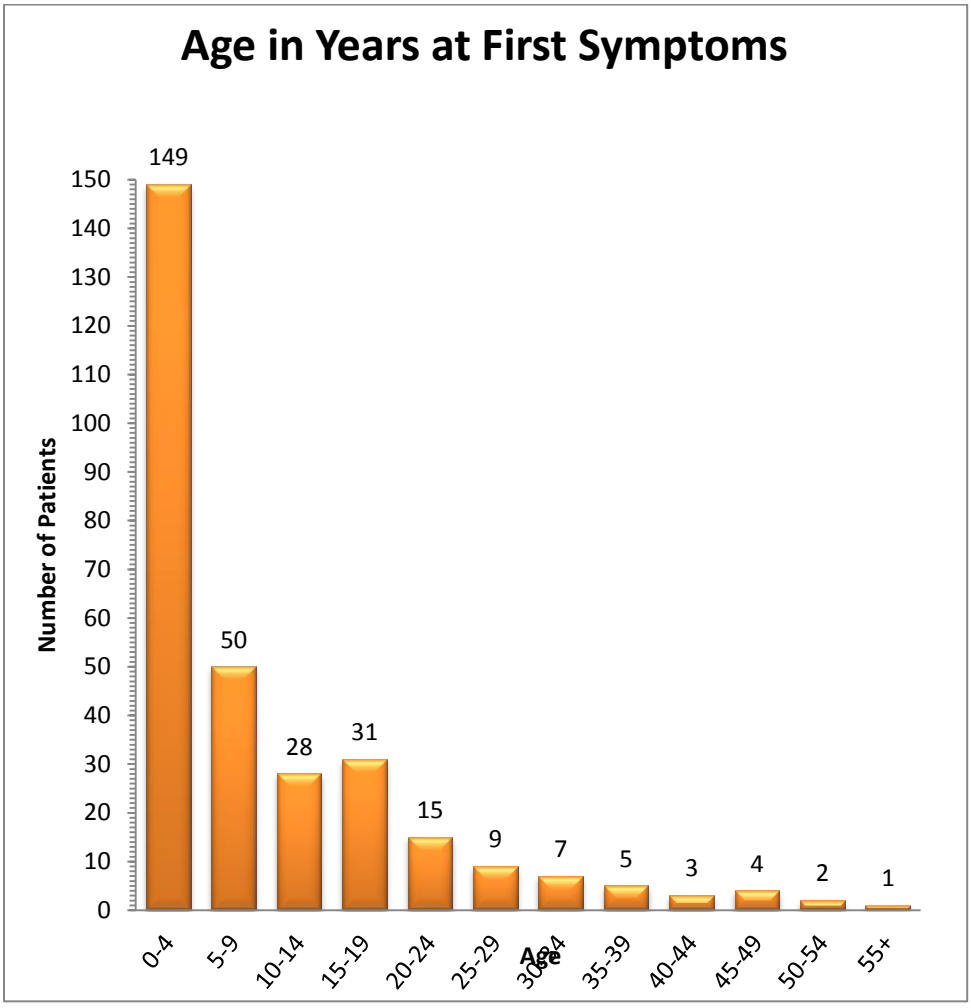
Of the 361 patients in the registry as of January 2013:

- **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**



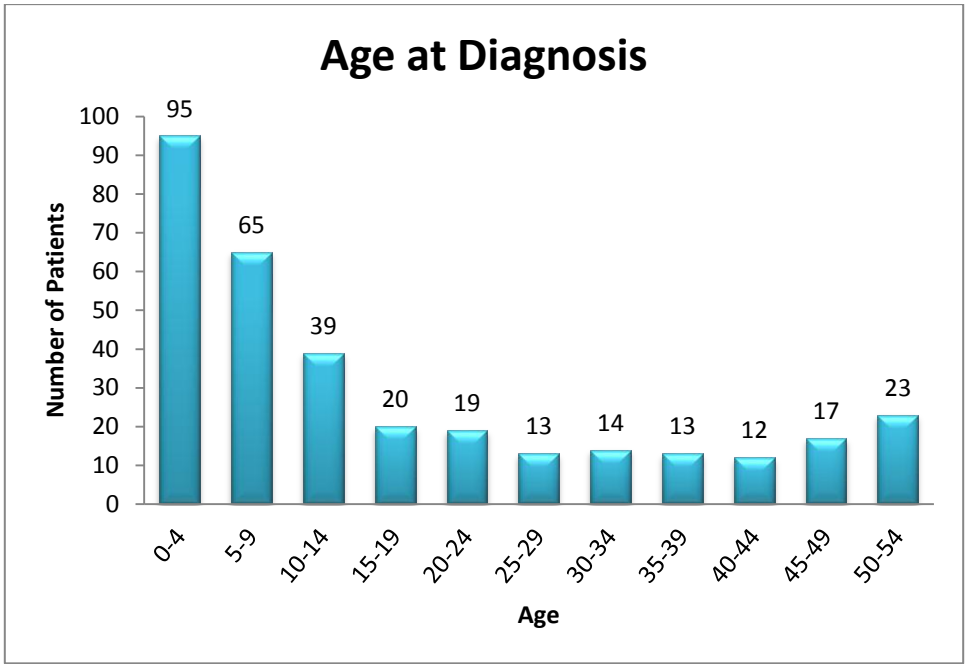
<u>Of the 361 patients in the registry as of January 2013:</u>

- **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**



<u>Of the 361 patients in the registry as of January 2013:</u>

- **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**