Cystine Stones: A rare and challenging group of Renal stones

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Introduction: Cystinuria is an autosomal recessive disorder in renal tubular and intestinal transport of dibasic amino acids, which results in increased urinary excretion of cystine, ornithine, lysine and arginine. Patients present with cystine calculi. Although the cystine calculi account for only about 1% to 2% of all renal stones but represent about 6% to 8% of all pediatric calculi. In this study we aimed to determine the frequency of cystine stones submitted for analysis at a clinical laboratory of a tertiary care hospital.

Methods: Design: A single center point prevalence study Setting: Department of Pathology & Laboratory Medicine in collaboration with Department of Surgery, AKU Calculi Analysis:
• Stones were analyzed by Fourier transformed infrared spectroscopy method by Nicolet iS5 Infrared spectrometer with the aid of an iD5 accessory, using Attenuated total reflection (Thermo Fisher Scientific Inc., USA)
• The FT-IR spectra of stones were matched against a library of spectra (Nicodom Renal Stones Library) to generate a report on stone components

Data Collection:
• Physical characteristics (colour, size, number) recorded
• Core and surface was analyzed separately for stones ≥2 cm
• The stones were categorized into pure (single component) and mixed stones
• Further classified as calcium oxalate monohydrate, calcium oxalate dihydrate, uric acid, carbonate apatite, ammonium urate, Struvite, cysteine and Xanthine stones

Figure 1: Frequency of Cystine Stones received

Total Stones received (n=1311)

Cystine Stones (n=45)

Other Stones (n=1266)

Results:
• Total 45 Cystine stones were tested with mean age [standard deviation (SD)] of the patients was 24 years (21.92), 73% (n=33) being male.

Table 1: Age and gender Distribution of Cystine stones

<table>
<thead>
<tr>
<th>Gender</th>
<th>Male</th>
<th>Female</th>
</tr>
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<tbody>
<tr>
<td>Age Category</td>
<td>≤16 years</td>
<td>&gt;16 years</td>
</tr>
<tr>
<td>Age (yrs)</td>
<td>10 (4)</td>
<td>30 (16)</td>
</tr>
<tr>
<td>Frequency (%)</td>
<td>8 (17)</td>
<td>25 (55)</td>
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</table>

• Average (SD) size of the stones was 1.5 cm (0.1-4.3). Only one patient was further tested for urine amino acid and reported as cystinuria

Figure 2: Distribution of Stones across Provinces of Pakistan

Conclusion:
This study shows that major composition of stones was calcium oxalate and geographical differences were also observed.
Cystine stones were common in male gender and commonly present in adult life.