Cystine lithiasis

Introduction
- **Prevalence**: 1-5% of all lithiases. Cystinuria is **Au recessive**.
- **Characteristics**: large yellow stones that are smooth, hard, and with variable radiopacity; bilateral in 75% of cases.
- **Normal cystinuria**: <30 mg/d.
- **Heterozygous hypercystinuria**: >200 mg/d; **homozygous hypercystinuria**: >400 mg/d.

Etiology
- **Reabsorption defect** in the proximal convoluted tubule of 4 **amino acids**, of which only cystine is insoluble and precipitates, forming stones. This pattern of aminoaciduria may be remembered with the acronym **COLA**:
  - Cystine
  - Ornithine
  - Lysine
  - Arginine

Diagnosis
- **Suspicious** if lithiasis appears before age 20, frequent recurrence, or family history.
- **Urinalysis**: network-like hexagonal crystals in the urine. Smell of **rotten eggs**.
- **Brand test**: qualitative colorimetric study of cyanide-nitroprusside + in urine. Detects cystinuria >75 mg/L.
- **Quantitative determination of cystine in 24 h urine. Cystinuria if >200 mg/24 h.**

Treatment
- **Fragmentation and extraction** with ESWL, percutaneous lithotripsy, or ureteroscopy. May be complemented by **chemolysis** with solutions of Acetylcystine or TromeThamine solution.
- **Protein restricted diet** (to reduce calcium and uricosuria and increase pH and citraturia).
- **Sodium restriction**: to less than 2 g/d. Reduces the excretion of cystine.
- **Diuresis** above 3 liters/day. Alkalinizing and citrus drinks are acceptable.
- **Alkalinization of the urine**: to maintain a pH between 6.5-8 (See chapter on Uric Acid Lithiasis). Also enhances the effect of chelating agents.

### Treatment Table

<table>
<thead>
<tr>
<th>Generic name</th>
<th>Brand name®</th>
<th>Dose</th>
<th>Duration</th>
</tr>
</thead>
<tbody>
<tr>
<td>Potassium citrate*</td>
<td>ACALKA, BLANEL</td>
<td>1 tab of 1 g/8 h</td>
<td>Tailored</td>
</tr>
<tr>
<td>Potassium citrate-Citric ac</td>
<td>URALYT URATO</td>
<td>2.5 g/8 h</td>
<td>Tailored</td>
</tr>
<tr>
<td>Sodium bicarbonate</td>
<td>BISODOL</td>
<td>1 g/6 h</td>
<td>Tailored</td>
</tr>
</tbody>
</table>

* ½ h after meals. In cases of CRF, Sodium bicarbonate is preferred (because it lacks potassium).

- **Cystine chelating drugs**: require blood count and biochemistry testing every 6 months.

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<tbody>
<tr>
<td>Acetazolamide*</td>
<td>DIAMOX</td>
<td>1 tab of 250 mg/night</td>
<td>Tailored</td>
</tr>
<tr>
<td>Captopril</td>
<td>CAPOTEN, CAPTOPRIL</td>
<td>1 tab of 50 mg/12 h</td>
<td>Tailored</td>
</tr>
<tr>
<td>D-Penicillamine**</td>
<td>CUPRIMINE</td>
<td>1 tab of 250 mg/d</td>
<td>Tailored</td>
</tr>
<tr>
<td>Merc propionylglycine</td>
<td>THIOLA</td>
<td>2 tab of 100 mg/6 h</td>
<td>Tailored</td>
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</tbody>
</table>

* Because it produces hypocitraturia, it may increase the risk of calcium phosphate lithiasis.
**Due to its toxicity; Pyridoxine (BENADON®) supplements of 1 tab of 300 mg/d is recommended.