STROKE-LIKE EVENT IN A 18-MONTH OLD AS INITIAL PRESENTATION OF ORNITHINE TRANSCARBAMYLASE DEFICIENCY

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BACKGROUND

- Ischemic stroke in children
  - annual incidence of 0.6 to 7.9/100,000 children
  - most commonly attributed to cardiac, vascular, hematologic or infectious aetiology

- Inherited metabolic disorders
  - some of them treatable
  - uncommon cause of stroke-like events

- Ornithine transcarbamylase deficiency (OTCD)
  - X-linked inborn error of metabolism of the urea cycle
  - can manifest at any age with episodes of hyperammonemia causing encephalopathy, and rarely metabolic stroke.
CLINICAL REPORT

18-month-old female
Growth P50
No selective refusal to food

Emergency Room: lethargy after a fall from a chair.
Past History: daily vomiting for up to 3 months
more recently somnolence and ataxia, with frequent falls.
Physical Examination: prostration, unstable posture, ataxia

Head CT scan: bilateral cortico-subcortical fronto-temporo-parietal hypodense areas, more extensive on the right

→ subacute stroke
## INITIAL INVESTIGATION

<table>
<thead>
<tr>
<th>Tests</th>
<th>Results</th>
</tr>
</thead>
<tbody>
<tr>
<td>Complete Blood Count, CRP</td>
<td>N</td>
</tr>
<tr>
<td>Coagulation tests, D-dimer</td>
<td>N</td>
</tr>
<tr>
<td>BUN, serum creatinine and electrolytes</td>
<td>N</td>
</tr>
<tr>
<td>Blood glucose, Lactate</td>
<td>N</td>
</tr>
<tr>
<td>Total protein, albumin</td>
<td>N</td>
</tr>
<tr>
<td>AST 160 U/L, ALT 232 U/L</td>
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<tr>
<td>Blood Gas: pH 7.35, HCO3 20 mmol/l</td>
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<tr>
<td>CSF</td>
<td></td>
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<tr>
<td>Homocysteine</td>
<td>N</td>
</tr>
<tr>
<td>Toxics (U) - negative</td>
<td></td>
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<tr>
<td>Thrombophilia screening - negative</td>
<td></td>
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<tr>
<td>Autoimmunity screening - negative</td>
<td></td>
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<tr>
<td>Head and Neck Doppler Ultrasound: Normal</td>
<td></td>
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<tr>
<td>Echocardiogram:</td>
<td>Normal</td>
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</table>
Brain MRI: the lesions had diffusion restriction only in the cortex and not in the white matter with normal MR angiography.

→ non-vascular insult, namely metabolic stroke.

No evidence of vascular, haematologic or infectious aetiology
Rapid neurological deterioration:
- Left brachial hemiparesis
- Reduced level of consciousness

Ammonia Levels (276 μmol/L)

Urea Cycle Disorder?

IMMEDIATE TREATMENT:
- High glucose and lipid infusion
- Ammonia scavengers
- Cessation of dietary protein

Rapid normalization of ammonia levels
- Progressive resolution of symptoms
- Partial seizure during recovery controlled with anti-epileptic therapy.

Aminoacids (P) and Organic acids (U)

<table>
<thead>
<tr>
<th>Aminoacid</th>
<th>Value</th>
<th>Change</th>
</tr>
</thead>
<tbody>
<tr>
<td>Glutamine</td>
<td>1258.9 uM</td>
<td>↑</td>
</tr>
<tr>
<td>Citruline</td>
<td>0.7 uM</td>
<td>↓</td>
</tr>
<tr>
<td>Orotic Acid</td>
<td>263.7 μmol/mmol creatinine</td>
<td>↑</td>
</tr>
</tbody>
</table>

6-MONTH FOLLOW-UP
- Hypoproteic diet + specific AA mixture + Arginine + Sodium Phenylbuturate
- Normal growth and slight psychomotor delay
- No further episodes of hyperamonemia
- Molecular genetic test is awaited.
• Our aim is to underline that urea cycle disorders should be thought in a patient with stroke-like episode especially if preceded of recurrent vomiting and ataxia.

• Prompt diagnosis and treatment is essential as duration and severity of hyperammonemia strongly correlates with brain damage.


THANK YOU FOR YOUR ATTENTION