P2-578
Cryptorchidism Is Commonly Observed in Allan Herndon Dudley Syndrome

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Background
- Allan-Herndon-Dudley syndrome (AHDS) is an X-linked mental retardation syndrome characterized by severe psychomotor retardation and pathognomonic thyroid parameters. Defects in monocarboxylate transporter 8 (MCT8), which facilitates thyroid hormone (TH) uptake and efflux across plasma membranes, have been linked to this disease.
- Schwartz et al. have reported the incidence of undescended testes as 8%.

Case 4
- 3y3m old boy
- Birth history
  - The patient was born by normal spontaneous vaginal delivery after a 41-week uncomplicated gestation. BW was 3254 g. There was no birth asphyxia and he received 48 h phototherapy for neonatal jaundice.
- Family history
  - No mental retardation or thyroid disorders
- Present history
  - Persistent head lag at age 5m prompted the primary care physician to refer the patient to our hospital. Thyroid function tests were: TSH 6.51 mIU/mL, FT4 0.69 ng/mL, FT3 8.29 pg/mL. Head MRI demonstrated delayed myelination. A p.G275R mutation in the SLC16A2 gene established the diagnosis of AHDS.
  - Physical examination and clinical course (tests)
    - On the first visit at age 6m, bilateral testes were 1-2 mL and palpable in the scrotum. At age 10m, ascending testes was noticed. Conservative observation was continued since the testes could be easily guided into the scrotum.
    - The testes became retractile by age 3y3m. The volume decreased to 1 mL and he was admitted for further examination at age 3y7m. Physical examination demonstrated bilateral testes in the inguinal canal. The position was determined as high scrotal and observation of testis was continued.

Endocrinological Data

<table>
<thead>
<tr>
<th>Age (y)</th>
<th>TSH (mIU/L)</th>
<th>FT4 (ng/mL)</th>
<th>FT3 (pg/mL)</th>
<th>LH (ng/mL)</th>
<th>FSH (ng/mL)</th>
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</thead>
<tbody>
<tr>
<td>3y3m</td>
<td>6.59</td>
<td>2.39</td>
<td>0.40</td>
<td>6.84</td>
<td>5.37</td>
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<tr>
<td>4y8m</td>
<td>4.89</td>
<td>7.5</td>
<td>0.7</td>
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<tr>
<td>5y1m</td>
<td>4.92</td>
<td>8.1</td>
<td>0.6</td>
<td>5.5</td>
<td>3.27</td>
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<tr>
<td>5y2m</td>
<td>8.35</td>
<td>7.1</td>
<td>0.5</td>
<td>5.5</td>
<td>3.27</td>
</tr>
<tr>
<td>5y3m</td>
<td>11.8</td>
<td>8.0</td>
<td>0.3</td>
<td>7.3</td>
<td>1.4</td>
</tr>
<tr>
<td>6y2m</td>
<td>10.8</td>
<td>5.0</td>
<td>0.3</td>
<td>5.7</td>
<td>0.7</td>
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<tr>
<td>6y4m</td>
<td>5.57</td>
<td>6.4</td>
<td>0.7</td>
<td>6.0</td>
<td>0.8</td>
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</table>

Objective
- To determine whether the prevalence of cryptorchidism is higher in severely affected AHDS patients than previously reported.

Patients and Methods
- A retrospective chart review of seven AHDS patients followed at our hospital was conducted.

Results
- Four out of seven AHDS patients had cryptorchidism. Another patient had severe retractile testes.

Conclusions
- 5/7 of the patients with severe AHDS presented cryptorchidism or retractile testes requiring orchiopexy.
- Sufficient TH transport may be necessary to sustain testicular descent.
- Early detection is needed to avoid the complications of cryptorchidism.