Urine steroid profiles: what can they do for me?

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Introduction
Assessment of urine steroid profiles (USPs) is a powerful test for investigating adrenal steroid disorders. Unlike most biochemical testing, USP analysis is very labour intensive, and full interpretation requires qualitative and quantitative assessment of the profile. For some disorders steroids must be identified manually, therefore adequate clinical information is essential to provide a complete report. Clinical indications for USP analysis are limited, typically including ambiguous genitalia, salt-losing states, virilisation, hypertension, and adrenal tumours. In 2017 we moved to a gas chromatograph-tandem mass spectrometer (GC-MS/MS), which has improved the specificity and throughput of the method. Here we present a review of USPs from the last 12 months to assess requesting patterns. We also present data from the new instrument examining the utility of steroid ratios for the diagnosis of late onset congenital adrenal hyperplasia due to 21-hydroxylase deficiency (LOCAH).

Requesting patterns
USP requests received between May 2017 and 2018 were extracted from the laboratory database. Clinical details were grouped according to possible condition and assessed for relevance to the following: 1) ambiguous genitalia, 2) salt-losing states, 3) adrenarche / virilisation, 4) hypertension, 5) adrenal tumour.

Requests categorised by age and gender are shown in Table 1. Clinical details received for these requests are shown in Figure 1. The majority of requests (~35%) were made to exclude LOCAH in the work up of adrenarche or polycystic ovarian syndrome, depending on the age of the patient.

Table 1 Total number of USP requests from May 2017-2018, categorised by age and gender.

<table>
<thead>
<tr>
<th>Age range</th>
<th>Female</th>
<th>Male</th>
<th>Unknown</th>
<th>Totals</th>
</tr>
</thead>
<tbody>
<tr>
<td>≤12 weeks</td>
<td>10</td>
<td>2</td>
<td>0</td>
<td>12</td>
</tr>
<tr>
<td>1-10 years</td>
<td>99</td>
<td>28</td>
<td>3</td>
<td>120</td>
</tr>
<tr>
<td>11-16 years</td>
<td>24</td>
<td>121</td>
<td>0</td>
<td>145</td>
</tr>
<tr>
<td>≥16 years</td>
<td>0</td>
<td>4</td>
<td>0</td>
<td>4</td>
</tr>
<tr>
<td>Totals</td>
<td>103</td>
<td>175</td>
<td>3</td>
<td>281</td>
</tr>
</tbody>
</table>

Most USP requests were made appropriately. However, 22% of requests were sent either without or with inappropriate clinical details. Inappropriate requests included GID, 11oxoP3, and were available for 54/151 cases (2 for 11oxoP3, 4 for 17P and 48 for P3).

Conclusions
1) Most urine steroid profile requests received in Glasgow are appropriate, but adequate clinical information is essential to provide helpful interpretation.

2) In agreement with Lucas-Herald et al., steroid ratios generated by the GC-MS/MS do not appear to offer significant advantages over steroid quantification in the diagnosis of LOCAH due to 21-hydroxylase deficiency. We are in the process of assessing the utility of steroid ratios in other clinical scenarios, in particular in adults for the differentiation of adrenal adenoma and carcinoma.

References

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