

RANGE OF URINARY STEROID METABOLITE RATIOS IN CHILDREN UNDERGOING INVESTIGATION FOR SUSPECTED DISORDERS OF STEROID SYNTHESIS

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BACKGROUND

Calculation of a urinary steroid metabolite ratio (uSMR) may be a useful method of improving diagnostic yield when investigating disorders of steroid hormone synthesis.

OBJECTIVE AND HYPOTHESIS

To investigate the range of uSMR in children with suspected disorders of steroid hormone synthesis. We hypothesise that there will be a difference in the ranges according to gender and age.

METHODS

Two ratios to assess 21 hydroxylase were calculated on steroid metabolite data analysed by GC-MS : **PT/(THE+THF+5 α THF)** and **17HP/(THE+THF+5 α THF)**. Urine samples were collected between 2008-2010 from 529 children who were undergoing investigations. To obtain reference data, samples were also analysed in 89 children with no background of endocrine concerns and who had a urine sample collected at presentation to the hospital with an acute illness.

RESULTS

A total of 529 patients who were being reviewed at the endocrine clinic were included.

- 186 (35%) of these were male
- The median age at test was 7.4yrs (1day-18yrs).
- The most common indication was the investigation of precocious puberty (35%)

A total of 89 patients with no background of endocrine concerns and who had a urine sample at presentation to hospital with an acute illness were included as the control group

- 36 (40%) of these were male
- The median age at the time of the test 3 years (range 1 month-11 years).

The number of cases with medians outside the range for the control groups is shown in Table 1. Figure 1 demonstrates the range of ratios for cases and controls.

Group	Total number controls	Total number cases	Number of cases >95 th centile PT/(THE+THF+5 α THF)	Number of cases >95 th centile 17HP/(THE+THF+5 α THF)	Number of cases >95 th centile both ratios
Girls <6 months	11	24	15	7	6
Boys <6 months	7	45	24	8	8
Girls 6 months-10 years	41	249	99	46	27
Boys 6 months-10 years	24	100	13	3	3
Girls >10 years	7	58	11	6	3
Boys >10 years	6	36	7	4	1

Table 2. Number of cases with medians outside the range for the control groups.

CONCLUSION

These novel data show that reference ranges for urinary steroid metabolite data need to be age matched. Most children with suspected disorders of steroid synthesis have a ratio which is within the reference range and the identification of outliers will lead to better targeting of genetic analyses.

PT/(THE+THF+5 α THF)



17HP/(THE+THF+5 α THF)

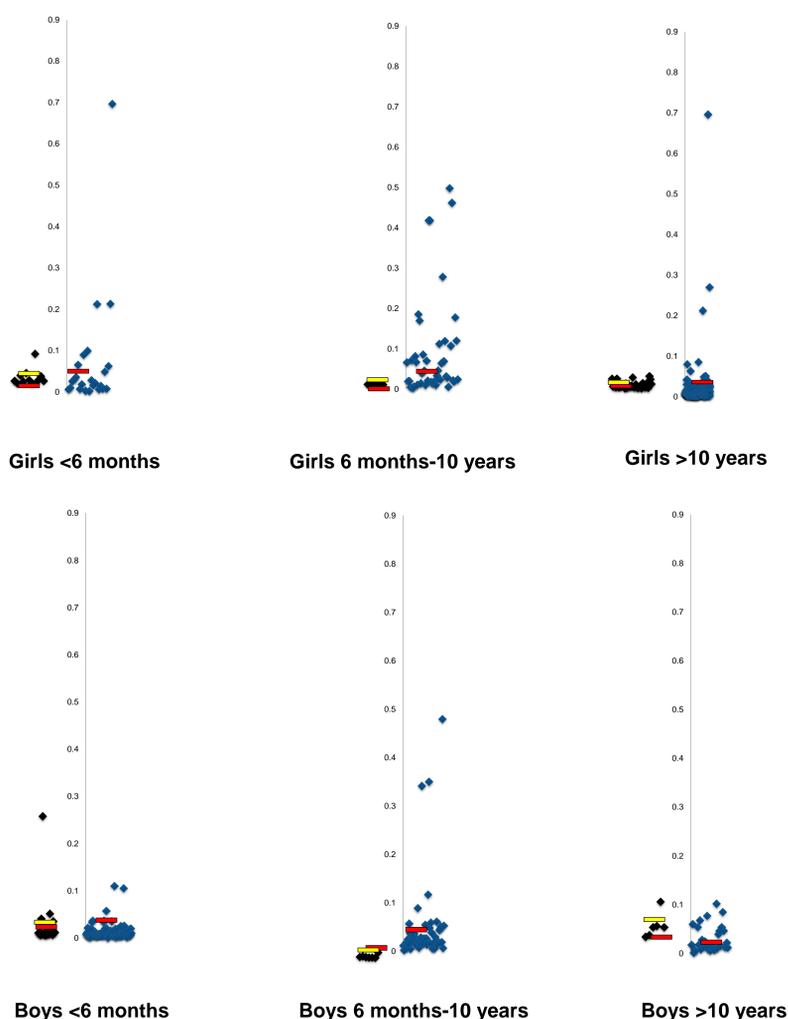


Figure 1. Ranges of urinary steroid ratios for 2 ratios 21 hydroxylase deficiency. The median is demonstrated by the red bar. The 95th centile is demonstrated by the yellow bar.

Abbreviations: 17HP: 17-hydroxypregnanolone, PT: pregnanetriolone, THE: tetrahydrocortisone, THF: tetrahydrocortisol.