TREATMENT IN CHILDREN WITH CONGENITAL ADRENAL HYPERPLASIA SHOULD BE INDIVIDUALISED, BUT PRECISELY HOW TO USE THE RESULTS FROM BIOCHEMICAL MARKERS IN THE CONTEXT OF BIOMETRIC MEASUREMENTS IS UNKNOWN. RECOMMENDATIONS OUTLINE A TARGET 17-OH PROGESTERONE (17OHP) OF 12-36 nmol/l, AND TO TARGET CONCENTRATIONS FOR ANDROSTENEDIONE (D4) IN THE NORMAL RANGE FOR AGE. WE AIMED TO STUDY THE VARIATIONS OF THESE BIOMARKERS IN PATIENTS FROM DIFFERENT CENTRES CONTRIBUTING TO THE INTERNATIONAL CONGENITAL ADRENAL HYPERPLASIA REGISTRY (I-CAH).

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**CONCLUSIONS**

Data from the I-CAH Registry show large variability in 17OHP and D4, with 17OHP most commonly above target range and D4 concentrations varying with age, and significant variability in 17OHP between centres. Concordance between the biomarkers suggests both have utility in disease monitoring. Further research is needed to establish optimal age- and sex-specific D4 targets in patients with CAH, to develop strategies for using these biomarkers in combination as monitoring tools, and to work towards standardising treatment to reduce unwarranted variation.

**METHOD**

Retrospective multi-centre study, including 21 centres (14 countries), analysed serum biomarker data from the International-CAH Registry. Data was available from 345 patients (52% females) under 18 years (median age 4.3 years IQR (Interquartile Range) 3.1 to 9.2 years) with classic 21-hydroxylase deficiency under follow-up between 2000 to 2020. The serum 17OHP and D4 concentrations of patients within different centres was summarised alongside their height, weight, and most recent dose of HC, and serum biomarkers compared between centres. Specific details about assays, laboratory techniques or timing of measurement of biomarkers in relation to administration of HC was not available. Statistical analysis was carried out in R.

**RESULTS**

Median 17OHP was within a target of 12-36 nmol/l in 15.9% of cases and 50.0% above this range. Differences in weight and dose are noted between age groups and thus direct comparisons between these cross-sectional cohorts should be interpreted with care. Comparing the pooled results from all centres in 5 year periods (2001-2005, 2006-2010, 2011-2015 and 2016-2020), there was no difference in median 17OHP or D4 concentrations in the last 20 years (p>0.05). There were differences in median 17OHP, ranging from a low median within centre of (2.0 nmol/l IQR 1.0 to 10.0) up to a high median within centre of 104.4 nmol/l IQR 46.1 to 225.73 (p<0.001) (Figure B). Correlation between 17OHP and D4 was stronger in patients under 12 years (Figure A). Correlation was similar in both sexes. Multiple regression of D4 against 17OHP covaried with age revealed consistent correlation between 17OHP and D4 (D4 = 0.608 x Age + 0.025 x 17OHP – 1.56, p<0.001, R2=0.29) showing age should be considered when interpreting these markers together.

**ACKNOWLEDGEMENTS**

This work would not be possible without the patients and the parents of the children with CAH whose data have been included in the I-CAH Registry.

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**REFERENCES**


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**INTERNATIONAL PRACTICE OF THERAPY MONITORING IN CONGENITAL ADRENAL HYPERPLASIA: REAL WORLD DATA FROM THE I-CAH REGISTRY**

**Aims**

To study the variations of 17OHP and D4 in patients under 18 years (median age 4.3 years IQR (Interquartile Range) 3.1 to 9.2 years) with classic 21-hydroxylase deficiency under follow-up between 2000 to 2020. To compare the results from all centres in 5 year periods (2001-2005, 2006-2010, 2011-2015 and 2016-2020), and to target concentrations for Androstenedione (D4) in the normal range for age.

**Conclusions**

Data from the I-CAH Registry show large variability in 17OHP and D4, with 17OHP most commonly above target range and D4 concentrations varying with age, and significant variability in 17OHP between centres. Concordance between the biomarkers suggests both have utility in disease monitoring. Further research is needed to establish optimal age- and sex-specific D4 targets in patients with CAH, to develop strategies for using these biomarkers in combination as monitoring tools, and to work towards standardising treatment to reduce unwarranted variation.

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