Pediatric Endocrinology

PEDIATRIC ENDOCRINOLOGY: ADRENAL, THYROID, AND GENETIC DISORDERS

Health Status of Children and Young Persons With Congenital Adrenal Hyperplasia in the United Kingdom: Results of a Multi-Center Cohort Study

Irina-Alexandra Bacila, MRCPCH, PGDipCH[1], Neil Lawrence, MBBCh, MRCPCH[1], Sundus Mahdi, BSc Psychology; MSc Health Psychology[2], Sabah Alvi, MBChB, MD[3], Timothy Cheetham, BSc, MBChB, MD, MRCPCH[4], Elizabeth Crowne, BSc, MB, CHb, MD[5], Urmi Das, MRCPCH, PGDip[6], Mehul Dattani, MBBS, DCH, FRCPCH, FRCP, MD[7], Justin H. Davies, MBBCh, MRCP, FRCPCH, MD[8], Evelien F. Gevers, MD PhD[9], Ruth Krone, MD FRCPCH[10], Andreas Kyriakou, MD[11], Leena Patel, MBBS, MD (India), FRCPCH, MHPEd, MD (Res)[12], Tabitha Randell, MBChB, MRCP, FRCPCH[13], Fiona Ryan, MBBCH, MD, DCH, MRCGP, FRCPCH[14], S Faisal Ahmed, MBChB, MD, FRCPCH[15], Nils Peter Krone, MD FRCPCH[16].

[1]The University of Sheffield, Sheffield, United Kingdom, [2]THE UNIVERSITY OF SHEFFIELD, SHEFFIELD, United Kingdom, [3]Leeds General Infirmaryl, Leeds, United Kingdom, [4]Great North Children's Hospital, Newcastle, United Kingdom, [5]Bristol Royal Hospital for Children, University Hospitals Bristol Foundation Trust, Bristol, United Kingdom, [6]Alder Hey Children's Hospital, Liverpool, United Kingdom, [7]Great Ormond Street Hospital, London, United Kingdom, [8]University Hospital Southampton, Southampton, United Kingdom, [9]Barts Health NHS Trust, Bedford, United Kingdom, [10]Birmingham Women's & Children's Hospital, Birmingham, United Kingdom, [11]Developmental Endocrinology Research Group, University of Glasgow, Glasgow, United Kingdom, [12]Paediatric Endocrine Service, Royal Manchester Children's Hospital, Manchester University NHS Foundation Trust, Manchester, United Kingdom, [13]Nottingham Children's Hospital, Nottingham, United Kingdom, [14]Oxford Children's Hospital, Oxford University Hospitals NHS Foundation Trust, Oxford, United Kingdom, [15]Royal Hospital for Children, Developmental Endocrinology Research Group, University of Glasgow, Glasgow, Scotland, United Kingdom, [16]University of Sheffield, Sheffield, United Kingdom.

Introduction: The association between congenital adrenal hyperplasia (CAH) and increased morbidity and mortality in adult life has been well established, however, limited knowledge exists regarding the onset of co-morbidities during childhood. **Objective:** To establish the health status of children and young persons (CYP) with CAH in the United Kingdom. **Methods:** This cross-sectional multi-center study involved 14 tertiary endocrine units across the United Kingdom. We recruited 107 patients aged 8-18 years with 21-hydroxylase deficiency and 83 matched controls. We collected and analyzed demographic, clinical, and metabolic data, as well as psychological questionnaires (Strengths and Difficulties [SDQ], Paediatric Quality of Life [PedsQL]and Self-Image Profile [SIP]). **Results:** The majority of patients (62.2%) were diagnosed within the first month of life, most commonly presenting with ambiguous genitalia (32.7%) or salt losing crisis (25.2%). After diagnosis, 37.3% of patients required admission for adrenal crisis, 11.2% presenting three or more episodes. Of the female patients, 57.6% had undergone urogenital examination under anesthesia and 35.5% had genital surgery. Most CAH patients received glucocorticoid (GC) replacement therapy with hydrocortisone (HC) (94.3%) and the rest with prednisolone, with a mean for relative GC doses of 13.3 (±3.7) mg/m^2 per day HC-equivalent. 76.6% of patients received treatment with fludrocortisone, with a mean dose of 105.0 (±50.2) µg/m^2 per day. Comparing height-Standard Deviation Score (SDS), patients under 12 years were taller ($p=0.011$) and patients aged 12-18 years shorter ($p=0.031$) than controls. Bone age was advanced in patients, with a mean difference from the chronological age of 1.9 (±2.2) years. CAH patients were more frequently overweight (26.4%) or obese (22.6%) compared to controls (10.8% and 10.8% respectively, $p<0.001$). Five patients had high blood pressure. A small number of patients had abnormal lipid profiles: raised total cholesterol (7%), low HDL (13%), raised LDL (4%) and triglycerides (15%). Insulin resistance as defined by HOMA-IR was found in 53.8% of patients with CAH, which was comparable to healthy controls (56.3%). 15.8% of patients had SDQ scores within the "high" and "very high" categories of concern. In the PedsQL questionnaires, 'school functioning' was the lowest scoring dimension with a median (IQR) of 70 (55 - 80), followed by 'emotional functioning' with a median score of 75 (65 - 85). The SIP scores were comparable to normative values, for all age and gender groups. **Conclusion:** Children with CAH have increased prevalence of growth and weight gain problems, metabolic co-morbidities, as well as reduced quality of life and mental wellbeing. There is a pressing need to optimize management and monitoring strategies in CYP with CAH order to improve long-term health outcomes.

Pediatric Endocrinology

PEDIATRIC ENDOCRINOLOGY: ADRENAL, THYROID, AND GENETIC DISORDERS

Health-Related Quality of Life in Cushing Disease: Discrepancy Between Parent and Child Reports

MARGARET F. KEIL, PhD, Adela Leahu, BS, Constantine A. Stratakis, MD, D(med)Sci, PhD(hc).
NIH/NICHD, BETHESDA, MD, USA.

Background: Previously we reported impairment in quality of life (QoL) measures in children with Cushing disease (CD) by parent proxy; however, little is known about QoL measures from the affected child's perspective. **Method:** Prospective study of 48 (26 females, Age:11.5±3.0 yr.) children diagnosed with CD. Prior to treatment, the Child Health Questionnaire (CHQ) was used to assess QoL by parent (proxy) and child report. **Results:** In children with active CD, there was a discrepancy between parent and child responses for mental health and self-esteem subscales. Child responses were significantly lower than parent responses (lower scores indicate poorer function) for mental health (56 ± 2 vs. 67 ± 2, p&lt0.001) and self-esteem (32.8 ±3 vs. 60.5 ±3, p&lt0.001). No significant differences were found between parent vs.child report for other subscale scores of the CHQ. **Conclusion:** This is the first study to compare child vs. parent report of health-related quality of life children with active CD. Parent reports underestimated the impact of CD on quality of life measures for mental health and self-esteem. It is important for clinicians to obtain information about health-related quality of life from the child's perspective in order