

## British Society for Paediatric Endocrinology and Diabetes and British Association for Paediatric Nephrology

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### EARLY MEDICAL TREATMENT OF GENDER DYSPHORIA: BASELINE CHARACTERISTICS OF A UK COHORT BEGINNING EARLY INTERVENTION

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**Aims** To describe characteristics of patients referred early (<16 yrs) medical treatment for gender dysphoria (GD). GD is a rare condition in which individuals experience clinically significant distress due to incongruence between their psychological perception of, and their natal assigned, sex.

**Methods** We collected data prospectively on all patients referred from May 2010–July 2014 for early pubertal suppression using gonadotropin – releasing hormone analogue (GnRHa) therapy.

**Results** 61 young people (34 natal males; 55.7%) were referred for early intervention to the national GD service endocrine liaison clinic at mean age of 13.1 years (range 9.8–15.3). All patients had a karyotype consistent with their natal sex. More natal males were in early puberty (32.4% Tanner 1/2; n = 11) than natal females (11.1% Tanner 1/2; n = 3).

Baseline endocrinology and physical examination were normal for natal sex in all patients. All females who had standard synacthen tests to exclude adrenal dysfunction (77.8%; n = 21) had normal cortisol and 17OHP. 38.2% (n = 13) males had low bone mineral density compared with 11.1% of females (n = 3).

50 patients (81.9%) elected to receive GnRHa following full explanation and informed consent at Tanner stage 3, following international guidelines. GnRHa could not be commenced immediately if pre-pubertal (10/61), having very low bone mineral density (3/61) or low body mass index (BMI) (2/61). All who began GnRHa achieved full gonadotropin suppression. No young people withdrew from GnRHa treatment in the first 2 years.

Many GPs were unwilling to prescribe GnRHa (56.0%; n = 28/50) therefore local hospitals (8.0%; n = 4) or the tertiary centre (36.0%; n = 18) issued prescriptions.

**Conclusion** Early medical intervention in GD with GnRH suppression of puberty is effective and well-tolerated. Assessment of growth, bone health and psychological outcomes will be important to assess the medium-and long-term safety and effectiveness of early intervention for GD.

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### THE RENAL STATUS OF THE UK JUVENILE – SLE COHORT

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Systemic lupus erythematosus (SLE) is a life-long severe disease; juvenile-onset (JSLE) patients experience more lupus nephritis

(LN) and progression to chronic kidney disease (CKD) occurs in some patients. Earlier detection of CKD in other conditions provides an opportunity to delay the rate of renal deterioration. Using a cross sectional analysis of participants recruited to the UK JSLE Cohort Study, the aim of this study was to assess the current renal function status and the presence of CKD associated factors (hypertension, proteinuria, hypercholesterolaemia, anaemia) in a cohort of JSLE patients.

The study cohort (n = 250) was aged 12.6 (10.2–14.3) years at diagnosis and 217 (87%) were female. The latest American College Rheumatology SLE score was 5 (4–7). The patients had JSLE for 3.8 (2.1–6.2) years, and a global British Isles Lupus assessment group index score of 2 (1–4). Seventy-nine (32%) patients had abnormal renal function; 43 (17%) had hyperfiltration (eGFR >140 ml/min/1.73m<sup>2</sup>), 24 (10%) had an eGFR 60–89 ml/min/1.73m<sup>2</sup>, 8 (3%) had an eGFR 30–59 ml/min/1.73m<sup>2</sup>, no patients had an eGFR 15–29 ml/min/1.73m<sup>2</sup> and 4 (2%) had an eGFR <15 ml/min/1.73m<sup>2</sup>. Hyperfiltration was associated with the presence of proteinuria (>500mg/day), seen in 32% (p = 0.03). With regards to CKD associated factors, hypertension was common in all renal function groups (26% of patients) and associated with reduced eGFR (p = 0.03) and proteinuria (p = 0.01). Hypercholesterolaemia only occurred in 10 patients (7%). Proteinuria correlated with the presence of hypertension (p = 0.014). Anaemia was not associated with renal function but with global disease activity.

This large UK wide inception cohort has demonstrated the burden of renal disease in JSLE. Recent studies have suggested renal hyperfiltration may have clinical significance, as it is independently associated with later renal function decline and cardiovascular disease in other conditions. This study may have identified JSLE patients at high-risk of CKD progression and it highlights three key findings; children with JSLE may have impaired renal function, CKD associated factors are common and shared care between Paediatric Rheumatology and Nephrology is required. Further confirmatory studies are required as early identification, combined with aggressive blood pressure management and reduction of proteinuria, may slow the rate of renal function decline in this population.

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### THE PATHWAY TO DIAGNOSIS OF TYPE 1 DIABETES IN CHILDREN: ANALYSIS OF FREE TEXT RESPONSES FROM A QUESTIONNAIRE STUDY

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**Aims** Distinguishing the rare child presenting with new onset type 1 diabetes (T1D) from the large number with similar symptoms and minor undifferentiated illness is challenging for both primary care physicians and families. This study analyses free text responses from a questionnaire completed by parents of children recently diagnosed with T1D to explore the parents' perspectives of the pathway to diagnosis of T1D in children.

**Methods** A questionnaire about the pathway from first symptom (s) to diagnosis was completed by the parent (s)/guardian (s) of children aged 1 month to 16 years diagnosed with T1D within the previous three months in the East of England. The free text questions asked what they felt symptoms were due to, what factors contributed to their decision to seek medical advice and whether they felt there was anything that delayed reaching the