on MDI and their average HbA1c values when on CSCI a year later. When repeated in 2019, there was an average improvement in HbA1c levels of 48% and at 24 months an average of 53% improvement.

Conclusion This data is utilised to demonstrate to staff and patients how CSCI alone is no guarantee for normalising poorly controlled HbA1C levels. We must continue to take a holistic approach to the counselling and management of our type 1 diabetes paediatric patients.

G426(P) DELAYED OR ABSENT? USE OF NEXT GENERATION SEQUENCING DIAGNOSTIC TOOLS IN A UK PUBERTY COHORT

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Background and Aim Isolated delayed puberty (DP) and congenital hypogonadotropic hypogonadism (cHH) are causes of severe delayed puberty that are almost clinically indistinguishable. Treatment is condition-specific and time-sensitive, yet existing methods to differentiate the two lack sensitivity and specificity. Genetic testing has been proposed as a potentially accurate diagnostic investigation. This project aimed to broaden our understanding of the genetic basis of severe delayed puberty to aid clinical diagnosis and prognosis of individual patients.

Methods Phenotype and genotype data of a UK cohort of 70 (probands = 24, relatives = 46) was studied. Whole exome sequencing was performed on probands and variants were filtered to identify candidate genes. Sanger sequencing was used to confirm variants and assess segregation with trait in pedigrees. Published literature, bioinformatics databases and prediction software tools were used to identify likely deleterious variants.

Results Following whole exome sequencing of 24 probands, 27 variants were identified in 21 candidate genes. 10 of these variants were found to be likely deleterious by prediction tools and 1 was proven to be definitively deleterious by previously published literature. 5 candidate genes had previously identified variants associated with cHH, 1 candidate gene had variants previously associated with isolated DP and another candidate gene had variants previously associated with both phenotypes. Clinical diagnoses was established in 46% of probands. After genetic testing, a further 38% had a likely pathogenic variant consistent with a diagnoses of cHH or isolated DP.

Conclusion A likely genetic diagnoses was established in 69% of probands with previously unclassified severe delayed puberty. Thus, identification of gene mutations known to cause cHH or isolated DP helps to clarify diagnoses. Genetic testing has the potential to allow time-sensitive, condition-specific treatment and future planning for individual patients. Though clinical significance of variants remains uncertain, *SEMA6A*, *SEMA3E* and *HS6ST1* were amongst candidate genes highlighted as potentially pathogenic providing plenty of scope for future study.

G427(P) ABSTRACT WITHDRAWN

G428(P) THE 50 MOST INFLUENTIAL STUDIES ON PAEDIATRIC DIABETIC KETOACIDOSIS: A BIBLIOMETRIC ANALYSIS

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Objectives Diabetic ketoacidosis (DKA) in paediatrics is a common condition and often the first presentation of type 1 diabetes mellitus in children. The number of citations a study receives can be used as a marker of academic influence, and can help to identify strong and weak areas of research within a particular field. We aimed to identify the 50 most influential studies relating to paediatric DKA by citation number, and analyse their characteristics.

Methods The Web of Science database was used to determine the studies that were most frequently cited. We searched for studies that included 'Diabetic ketoacidosis' or 'DKA' along with 'paediatric' or 'children' or 'child' or 'infant' in the title. Data extracted included publication year, article type and focus, publication journal, institution and country, level of evidence (LOE) and total citation number. Published guidelines were excluded from the LOE rankings. Citation density was also calculated as number of citations per year since publication.

Results Our search returned 410 studies. The top 50 studies were published between 1976 to 2014. The most common study topics were epidemiology and pathophysiology, with many studies relating to cerebral oedema (n=19). The mean number of citations was 74.7 (standard deviation 60.3, range 28–375). These were published in 21 journals, led by *Pediatric Diabetes* (n=9). They originated from 9 different countries, with institutions from the United States of America publishing the majority (n=30), and the University of California, Davis publishing the most studies (n=7). One RCT was identified, 39 non-randomised clinics studies were included. The remainder included reviews, basic science studies and guidelines. 10 studies were classified as LOE 1, and 11 studies as LOE 4.

Conclusions The 50 top cited studies on paediatric DKA relate mostly to epidemiology and pathophysiology. Cerebral oedema in paediatric DKA patients has received increased attention by researchers and institutions. LOE was low, with only one RCT identified. Though these studies are the most influential in the field, perhaps the evidence base for this field would benefit from further high quality trials.

G429(P) DIAGNOSIS OF DIABETES MELLITUS IN CHILDREN, INCLUDING FACTORS LEADING TO DELAYED DIAGNOSIS

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Aims To identify patient demographics, presentation and contributing factors towards delay in new diagnosis of diabetes mellitus in children, and to compare this to results from a similar audit in 2013.

Methods A retrospective audit of electronic records of all patients identified from the local paediatrics diabetes caseload database with new diagnoses between 1st January and 31st December 2018. Inclusion criteria included age at diagnosis

(0–16yo), place of diagnosis (local hospital), and new diagnosis.

Results 21 patients were identified within the database; four were excluded due to diagnosis in other country or lack of notes. 9 females and 8 males were included, with a mean age of 8 years and a median duration of presenting symptoms of 21 days. 82% were referred to the paediatric emergency department by their general practitioner (GP). 82% reported polyuria, 76% reported polydipsia, 29% reported fatigue and weight loss, with other presenting symptoms including nocturia and high capillary blood glucose measured at home. 76% had a family history of diabetes or autoimmune disease. 82% presented with hyperglycaemia, and 18% in diabetic ketoacidosis. Mean presenting capillary blood glucose was 24 mmol/L with a mean HbA1c of 106 mmol/mol. Only 2 patients (12%) had urine dipstick results documented on their hospital discharge summary. 16 subjects had a new diagnosis of type 1 diabetes and 1 had a new diagnosis of type 2 diabetes. Compared to previous data from 2013, in which 50% of patients had a delay in diagnosis, 3 patients (18%) had delayed diagnosis of diabetes. All of this group were seen prior to diagnosis by their GP, with a median duration of delay of 3 days.

Conclusion Delay in diagnosis of diabetes in children appears to have reduced in the past 5 years in the local area. However, practice must continue to improve to prevent unacceptable delay. This requires education to improve symptom recognition, of both local healthcare professionals, and also of adult patients with autoimmune disease, in view of the high incidence of diabetes in children with a family history.

G430(P) USING THE HOSPITAL SCHOOL AS A RESOURCE TO WIDEN THE SCOPE OF CONVERSATION DURING CLINICAL DIABETES CONSULTATIONS

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Introduction Exploring the impact of diabetes on the wider life of ‘school examination aged young people’ is thought to be really important. We hoped that doing this ‘Me First’ approach in a demonstrative way would encourage greater self efficiency in the management of both their diabetes and everyday life. At this age, preparation and completion of examinations is one of the most important elements of a young persons life, so it made sense to involve the Hospital School as part of our clinical consultations.

Method We introduced a ‘meet and greet’ system, which involved the young person being greeted on arrival in clinic by a hospital school teacher. They sat down immediately with the teacher, discussed school matters, also completing an ‘All About Me’ form, with time to talk over their choices. The ‘All About Me’ form gathers personal information such as family support, likes, dislikes, goals, ambitions, social and educational needs and preferences. Young People were also given specific information regarding extra time and special consideration for exams. They then went on to see the diabetes team as usual. This method also eliminates ‘the waiting aspect’ to clinic.

The young people and staff were then asked to give feedback regarding the new approach to consultations.

Results

- Young people and staff reported the ‘All About Me’ form, gave improved depth and quality of conversation around their diabetes, putting their needs at the centre of the clinic visit from the very start.
- 100% of the diabetes team felt this led to easier flow of conversation.
- 100% of the young people were not aware that access arrangements and special consideration could be applied for on the grounds of having diabetes.
- 100% of the young people had experienced diabetes related issues around completing examinations.
- 100% of the young people requested their schools to be contacted and informed regarding access arrangements.

Conclusion Discussion around school and using the ‘All About Me’ form, clearly placed young people at the heart of the consultation, ‘Me First’ approach, and provided an opportunity for them to discuss what matters to them as well as being more able to articulate their own goals.

Young people are underinformed regarding school exam access arrangements and are eager to embrace this, consenting to their schools being informed.

G431(P) GENDER DYSPHORIA – A DESCRIPTION OF THE CHANGES IN PREVALENCE, DEMOGRAPHICS AND THE CLINICAL CARE PROVIDED BY A PAEDIATRIC ENDOCRINOLOGY DEPARTMENT

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Aim To describe the trends in prevalence, clinical characteristics and clinic journey of young people (YP) attending the paediatric endocrine service, because of Gender Dysphoria (GD).

Methods Electronic medical records of YP attending the paediatric endocrine service, between 2011–2018, were reviewed.

Results Overall, 74 YP [49 (66%) birth-assigned females and 25 (34%) birth-assigned males] were assessed by paediatric endocrinology in 2011–2018. Forty-six (62%) were referred in the 2-year period 2017–2018. Median age (range) at initial assessment was 14.6 yrs (8.8,17.5) and remained unchanged during the study period ($p=0.862$). Median age (range) at initial assessment was 14.3 yrs (8.8,15.8) for birth-assigned females and 15.0 yrs (9.8,17.5) for birth-assigned males ($p=0.096$). There was a 4-fold increase in the number of YP attending paediatric endocrinology from 2015 ($n=15$) to 2018 ($n=60$). Overall, 55/74 (36 birth-assigned females, 19 birth-assigned males) were eligible for referral for fertility preservation, of which 18 (33%) (12 birth-assigned females, 6 birth-assigned males, $p=1.0$) sought a formal referral to Fertility service. One (8%) birth-assigned female and 5 (83%) birth-assigned males completed fertility preservation ($p=0.004$). Sixty-five (88%) of those attended [44 (90%) birth-assigned females and 21 (84%) birth-assigned males] were commenced on gonadotrophin-releasing hormone analogue (GnRHa) for puberty suppression. Of those, 46 (71%) were commenced on GnRHa in