## GP137 A COMPARATIVE ANALYSIS OF CHILDREN AGED 4–13 YEARS WITH TYPE I DIABETES WHO WERE INVITED TO TAKE PART IN THE 'TEAM TYPE I' INITIATIVE

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**Aims** To bring together a group of children with Type I Diabetes Mellitus (T1DM), with their siblings and parents, to participate in a group fun run. Alleviate any barriers to exercise they may previously have had by setting up a hypoglycaemia treatment table and having the option of chaperones to run with the children. Gather information about diabetes care and exercise by means of survey.

**Methods** Flyers advertising the initiative were posted to all eligible children. Paper surveys were given to those who participated, and phone surveys were conducted for those who didn't. Results were tabulated.

**Results** Runners (R) n=16. Non-Runners (NR) n=14. The mean age (9.4 years) and HbA1c (R: 8.061% or 64.6 mmol/mol, NR: 8.077% or 64.8 mmol/mol) levels for both groups were similar. Majority of both groups used continuous subcutaneous insulin therapy (R: 62.5%, NR: 64.3%). Interestingly, although R's had experienced a greater number of hypoglycaemic episodes in the previous two weeks (Mean R: 6.875, 0–20. NR: 5.43, 2–10), 25% of R's reported the fear of hypoglycaemia would stop them from exercising, compared with 35.71% of NR's. This could be explained by the fact that NR's had experienced more severe hypoglycaemic episodes in the previous year (R: 1, NR: 3). Blood glucose monitoring by finger prick was more frequent amongst the R's with majority testing 10+ times per day. 71.43% of NR's and only 43.75% of R's reported having zero friends with T1DM. 100% of R's had taken part in a group fun-run before whereas 92.86% of NR's had not.

**Conclusion** Demographics and relative diabetes control is similar across groups. Our results highlight differences in the attitudes and actions around hypoglycaemia and exercise and also in the support networks available to these children. The results from our survey will allow us to continue this initiative annually and future surveys will allow us to further study motivations and barriers to exercise in the paediatric population with T1DM.

## GP138 RESILIENCE ENHANCE THE PROTECTIVE IMPACT OF FAMILY FUNCTIONING ON DIABETES DISTRESS IN YOUTH WITH TYPE 1 DIABETES

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**Objectives** a) to explore the effect of family functioning on diabetes distress among adolescents with type 1 diabetes(T1D); and b) to examine whether resilience mediates the relationship between family functioning and diabetes distress.

**Methods** Youth with T1D recruited from a national endocrine center of a public hospital in China from May 2017 to October 2018. A total of 189 participants (aged 8–24 years) completed the survey about their resilience, family adaptability and cohesion, diabetes distress and provided demographic and clinical information. The moderation analyse was preformed to determine whether the resilience strengthens the protective impact of family adaptability and cohesion on diabetes distress. The simple slopes analyse was used to probe significant interactions.

**Results** The mean score of diabetes distress was 29.58±22.09 with 31.7% of patients having severe diabetes distress. Multivariate linear regression analyses indicated that resilience enhanced the association that high family functioning had with low diabetes distress ($\beta$ =-0.22,$t$=-0.318, $P$=0.002). However, simple slopes found that benefits of high resilience for lower diabetes distress was only apparent in the context of low family adaptability and cohesion($\beta$ =-0.941, $t$=-4.090, $P$ =0.001).

**Conclusions** Many youth with type 1 diabetes reported severe diabetes distress which was associated with poor glycemic control and decreased quality of life. The finding of study suggest that family-based interventions which considered resilience factors are promising for youth with high diabetes distress especially for those have poor family functioning.

## GP139 THE INCIDENCE OF TRANSIENT PSEUDOHYPOALDOSTERONISM IN INFANCY IN IRELAND: A PROSPECTIVE WHOLE ISLAND SURVEILLANCE STUDY

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**Aim** To review the clinical features, presentation, investigations undertaken, and outcome of infantile salt-wasting presenting in the setting of urinary tract infection (UTI) and/or urinary tract malformation (UTM) over a two-year surveillance period on the island of Ireland. To estimate a population incidence based on the results and to make recommendations on the approach to management of this condition.

**Methods** A two-year (2013–14) prospective surveillance undertaken for the island of Ireland via the Irish and Ulster Paediatric Surveillance Units. Monthly-prepaid postcards were circulated to Consultant Paediatricians (n = 260) at all Paediatric Units on the island of Ireland.

Infants under one year of age presenting for the first time with hyponatremia (serum sodium < 130 mmol/L) and/or hyperkalemia (serum potassium > 5.0 mmol/L) associated with urosepsis/UTM were included.

**Results** Seven patients (six male), all aged younger than five months (3 weeks to 20 weeks) were reported during the study period. All had culture-proven UTI and five (71%) also had an underlying UTM (one diagnosed antenatally). Four (57%) patients had a documented elevated serum aldosterone supporting secondary pseudohypoaldosteronism (PHA) as the underlying diagnosis. Data on aldosterone was not reported in the other three patients but clinical features were suggestive of secondary PHA. All had an excellent outcome with full resolution of the electrolyte disturbance. No cases of primary PHA were submitted during the surveillance period. The estimated

incidence for the Irish population of transient pseudohypoaldosteronism was 1 per 13,200 live births per year for the study duration.

**Conclusions** Salt-wasting is a rare complication of UTI, especially if associated with underlying UTM. There is a similar annual incidence rate to the previously reported incidence of congenital adrenal hyperplasia in Ireland. Boys appear to be at particular risk. Prognosis is good if the condition is recognised and managed promptly.

## GP140 CYCLICAL CUSHING'S SYNDROME: A DIAGNOSTIC CHALLENGE

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**Aims** Cyclic Cushing's syndrome is an uncommon disorder, defined by intermittent episodes of excess cortisol secretion. These episodes occur sporadically. The fluctuating clinical picture and conflicting biochemical findings make Cyclic Cushing's syndrome challenging to diagnosis. We report a case of Cyclic Cushing's syndrome is a 6 year old boy and discuss the challenges in diagnosis.

**Methods** A detailed chart review was performed. Data extracted from the medical records included presenting complaint, disease progression, laboratory results, imaging and clinical measurements.

**Results** At 4 years of age the patient presented with a two week history of rapid weight gain, increased appetite, lethargy, polydipsia and polyuria. The child has a background history of speech delay, obesity (weight 29 kg, >99.6$^{th}$centile, BMI 23.3 kg/m$^2$), macrocephaly (OFC 56cm, > 97$^{th}$centile), and facial freckling. During an in-patient stay, six hourly serum cortisol levels taken over a 48 hour period followed by a dexamethasone suppression test and a 24 hour urinary cortisol collection failed to support a diagnosis of Cushing's Syndrome. His significant facial freckling -with lip sparing and no mucosal involvement - prompted a Clinical Genetics referral. A diagnosis of Carney Complex (CNC) with a mutation in the PRKAR1A gene was made.

The patient continued to have episodes that would suggest episodic hyper-secretion of cortisol. Each episode lasted 3–4 weeks and then resolved. Parents reported 2 episodes in 2016, 1 episode in 2017 and 1 episode in 2018. In early 2018, the patient was admitted to hospital during an acute episode. A diagnosis of Cyclical Cushing's syndrome was confirmed by very elevated serum cortisol levels, elevated 24 hour urine free-cortisols, failure to suppress to dexamethasone and a very suppressed ACTH level during this admission. MRI and CT of abdomen however failed to reveal any adrenal lesions. Following discussions at multidisciplinary team meetings and with colleagues in adult Endocrinology, a decision was made to proceed with a bilateral adrenalectomy. The patient tolerated the procedure well. His adrenal histology was consistent with subtle changes suggestive of a mild Primary pigmented nodular adrenocortical disease (PPNAD) picture. Following adrenalectomy his symptoms have completely resolved but he will require lifelong Hydrocortisone and Fludrocortisone replacement.

**Conclusion** Our patient has a background history of CNC and PPNAD which has been linked in very occasional cases with Cyclical Cushing's syndrome. PPNAD is the most common endocrine manifestation of CNC. This case report highlights the difficulty in diagnosing Cyclic Cushing's syndrome.

## GP141 POSSIBLE AGGRAVATION OF DESMOID TUMOURS WITH PRIMARY OVARIAN FAILURE TREATMENT

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**Introduction** Chromosomal abnormalities are a common cause of primary ovarian failure in adolescents. Familial adenomatous polyposis (FAP) is an autosomal dominant predisposition to developing colonic polyposis arising from a germline mutation in the APC gene. Desmoid tumours (DT), otherwise known as 'deep fibromatosis', are locally invasive tumours that do not metastasize. DT develop in between 5–30% of FAP carriers and are the second leading cause of death after colorectal carcinoma. Inductions of DT growth in both pregnancy and during oestrogen therapy have been reported. Selective oestrogen receptor modulators, including tamoxifen, are currently being utilised as a therapeutic agent for these tumours.

**Case description** A 14 year old girl was referred due to secondary amenorrhea and raised FSH levels. She entered puberty spontaneously, reached menarche at 13 years of age and had a regular 28 day menstrual cycle. She subsequently became amenorrheic 5 months later.

The index case was born at term by spontaneous uncomplicated vaginal delivery with a birth weight of 3.6 kg. Subsequent failure to meet development milestones led to a hearing assessment at 9 months of age which diagnosed bilateral sensorineural hearing loss and bilateral cochlear implants were inserted. Genetics at that stage demonstrated a *de novo* chromosomal translocation involving the×chromosome and chromosome 15 (46X translocation(X;15)(q13;q13).ishXq13 (Xist×2)).

Her father was subsequently found to be an FAP carrier and she is confirmed positive for this mutation. She has had annual surveillance colonoscopies; the most recent in June 2018 identified two adenomatous polyps. At 12 years of age she developed desmoid tumours, one located in the submandibular area and the other in the periumbilical area.

Investigations confirmed primary ovarian failure with undetectable oestradiol in the presence of elevated gonadotropins, normal androgens and low AMH levels. All other investigations were normal.

**Discussion** Oestrogen therapy is the mainstay of treatment in primary gonadal failure however, exogenous oestrogen is a risk factor in the exacerbation of desmoid tumour growth which could be life limiting. The family and their medical team need to strike a balance enabling optimisation of bone health without increased morbidity from tumour growth. Options were explored with the patient and her family with the ultimate decision to refrain from using exogenous sex hormone therapy with optimization of bone health. This is a complex case that poses therapeutic challenges in management and treatment goals of primary gonadal failure in the setting of desmoid tumours.