A COMPARATIVE ANALYSIS OF CHILDREN AGED 4–13 YEARS WITH TYPE 1 DIABETES WHO WERE INVITED TO TAKE PART IN THE ‘TEAM TYPE 1’ INITIATIVE

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Aims To bring together a group of children with Type 1 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 had 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.

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 analysis was preformed to determine whether the resilience strengthens the protective impact of family adaptability and cohesion on diabetes distress. The simple slopes analysis was used to probe significant interactions.

Results The mean score of diabetes distress was 29.58±22.09 with 31.70% 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 (β = -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(β = -0.941, t=-4.090, P =0.001).

Conclusions Many youth with type 1 diabetes reported severe diabetes distress which was associated with poor glycomic 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.

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 sepsis/UTI 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 pseudohypoa-
dosteroneism was 1 per 13,200 live births per year for the
study duration.

**Conclusions** Salt-wasting is a rare complication of UTI, espe-
cially if associated with underlying UTM. There is a similar
annual incidence rate to the previously reported incidence of
genital 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 pic-
ture and conflicting biochemical findings make Cyclic Cus-
hing’s syndrome challenging to diagnosis. We report a case of
Cyclic Cushing’s syndrome in 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 com-
plaint, 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, >99th centile, BMI
23.3 kg/m²), macrocephaly (OFCC 56cm, > 97th centile), and
facial freckling. During an in-patient stay, six hourly serum
cortisol levels taken over a 48 hour period followed by a dex-
amethasone suppression test and a 24 hour urinary cortisol
collection failed to support a diagnosis of Cushing’s Syn-
drome. 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 Cyclic Cushing’s syndrome was confirmed by
very elevated serum cortisol levels, elevated 24 hour urine
free-cortisol, 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. Follow-
ing 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
adenocortical disease (PPNAD) picture. Following adrena-
lectomy 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 Cyclic Cushing’s syndrome. PPNAD is the most com-
mon 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 adenoma-
tous polyposis (FAP) is an autosomal dominant predisposition
to developing colonic polyposis arising from a germline muta-
tion 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 color-
cectal carcinoma. Inductions of DT growth in both pregnancy
and during oestrogen therapy have been reported. Selective
oestrogen receptor modulators, including tamoxifen, are cur-
cently being utilised as a therapeutic agent for these tumours.

**Case description** A 14 year old girl was referred due to secon-
dary amenorrhoea 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
amenorrhoeic 5 months later.

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

Her father was subsequently found to be a 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 subman-
dibular area and the other in the periumbilical area.

Investigations confirmed primary ovarian failure with unde-
tectable oestriol in the presence of elevated gonadotropins,
normal androgens and low AMH levels. All other investiga-
tions 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 hor-
mone 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.