results we used the method Wilcoxon F. Results: At the beginning of the treatment the level of glyhemoglobin was 7.8% and the sleep duration was 6.4 hours.The included patients were treated by cognitive behavioral therapy for 6 months without changing other kinds of therapy. The sleep duration significantly(P=-0.35) enlarged until 7.5±0.2 hours and the level of glyhemoglobin was lowered by 0.4% significantly (P=0.47). The improving of sleep duration can successfully reduce the level of glyhemoglobin in diabetes mellitus type 2 without any intervention at glycemia and could contribute to the prevention of diabetes complications.

Bone and Mineral Metabolism

BONE DISEASE FROM BENCH TO BEDSIDE

Obesity and Impaired Glucose Metabolism in Adult Patients with X-Linked Hypophosphatemia

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SUN-336

Background: X-linked hypophosphatemia (XLH) is characterized by increased activity of circulating FGF23 resulting in renal phosphate wasting and abnormal bone mineralization. Epidemiologic studies suggest a relationship between FGF23, obesity and metabolic syndrome. However, the prevalence of metabolic complications in adult XLH patients is not known.

Study design: We conducted a prospective cohort study (CNIL 2171036 v 0) in a single tertiary referral center.

Subjects: Patients were adult subjects with XLH, defined as hypophosphatemia due to renal phosphate wasting with either documented *PHEX* mutation (92%) and/or family history of rickets. Healthy controls were selected among adult participants of the AcroCut cohort and matched for sex, age and body mass index (BMI).

Methods: Prevalence of obesity and diabetes in patients was compared with the general French population (ObEpi 2012; Bonaldi C. *et al.*, Bulletin épidemiologique hebdomadaire 2016) and prevalence of glucose intolerance in patients, assessed by standard 75-g OGTT, was compared to matched controls. Resistance to insulin was evaluated with HOMA-IR index.

Results: 107 patients (79 women and 28 men) were recruited. Median age at evaluation was 35.5 years (range 16.9-74.2) and median BMI was 25 kg/m^2 (range 18-48). Thirty-eight (35.5%) patients were overweight (29 women) and 22 (20.5%) patients were obese (16 women). Distribution of normal weight, overweight and obesity in adult XLH patients differed from that seen in the general French population. We observed an excess of overweight and obesity (+20%, 95% CI [+6; +33], *P*=0.013) in comparison to general population, especially in younger age categories (under 45 years of age). Three (2.8%) out of the 107 patients were treated for diabetes which did not differ from the expected frequency of type 2 diabetes of 5% in the French population. Twelve (13%) out of 90 patients with available OGTT were glucose intolerant or diabetic compared to ten (12%) of the 82 matched controls (*P*=0.995). Decreased insulin sensitivity assessed by HOMA-IR was found in fourteen (15.6%) adult XLH patients. Comparison of insulin and glucose curves obtained during the OGTT in patients and matched controls are planned to analyze insulin sensitivity in both populations.

Conclusion: Adult XLH patients are prone to develop overweight and obesity, particularly adolescents and young adults. This excess of weight does not seem to result in increased prevalence of metabolic disorders. Insulin sensitivity, yet, needs to be further evaluated in these patients and compared to appropriate controls. FGF23 concentrations will also be assessed to search for determinants of insulin sensitivity in patients. Lifestyle recommendations to prevent obesity, promoting physical activity, are thus essential in the management of XLH patients.

Pediatric Endocrinology

PEDIATRIC ENDOCRINE CASE REPORTS I

Unusual Case of Short Stature and Poor Growth in Childhood

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SAT-060

Unusual Case of Short Stature and Poor Growth in Childhood

Background: Néstor-Guillermo progeria syndrome (NGPS; OMIM 614008) is caused by biallelic pathogenic variants in *BANF1* (barrier-to-autointegration factor 1) on chromosome 11q13. It characterized by early onset and slow progression of symptoms including poor growth, lipoatrophy, pseudo-senile facial appearance, and normal cognitive development. Two adult patients have been reported. This is the first reported case of a child with NGPS who presented to endocrine clinic with failure to grow.

Clinical Case: Two year, 8 month old Hispanic female born at 40 weeks gestation with birth weight 3.5 kg. At 1 year, she had short stature, poor weight gain, and thinning hair. There were no developmental concerns. Family history was remarkable for consanguinity. At presentation, her weight was 8.5 kilograms) and height 80 centimeters (both <1st

percentile) and head circumference 45.5 centimeters (3rd percentile). Hair was sparse and fine with large areas of scalp alopecia. She had a small face with overhanging brow ridge, flattened midface, narrow nose, small mouth and bilateral lower eyelid ectropion. Fingers were shortened with thickened knuckles, widened fingertips, and distally set nails. Skin was tight throughout, particularly notable on the legs and hands with light discoloration of skin over the hand joints and reticulated dark macules over the lower abdomen. Her cardiac, respiratory, abdominal, genitourinary, neuro and joint examinations were unremarkable. Routine labwork was normal. Her bone age was normal at 2 year and 7 months but there was hypoplasia of the distal phalanges. Full skeletal survey revealed small mandible, thinning of the cranial vault, apparent crowding of the teeth, short stature, acroosteolysis-like changes involving the distal phalanges most evident in the hands, pointed distal phalanx of the great toes, and resorption of the distal clavicles. Her echocardiogram was normal. Sequencing and deletion/duplication analysis of *LMNA* was not diagnostic. Trio-based whole exome sequencing (WES) was performed after obtaining informed consent. WES revealed homozygosity for a pathogenic missense variant in *BANF1* c.34G>A (p.Ala12Thr) inherited from each of the unaffected parents. **Conclusion:** Progeria syndromes are unusual but diagnosable causes of failure to grow and can be diagnosed based on clinical suspicion. This patient represents the first child reported with NGPS.

Reproductive Endocrinology

MALE REPRODUCTIVE CASE REPORTS

Tetraorchidism: A Rare Type of Polyorchidism.

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SUN-030

Polyorchidism is defined as the presence of two or more testis. We report an interesting case of Tetraorchidism an extremely rare type of Polyorchidism in a 14-year-old boy with short stature due to Growth hormone deficiency. An extensive review of literature yielded only 200 case reports of Polyorchidism [1]. Most of these are case reports on triorchidism (3 testicles). There has been only 9 case reports of patients with (tetraorchidism) 4 testicles [2]. Most of them has been reported as incidental findings in adults.

This case is being reported due to its rarity. Here we elaborate on clinical presentation of Polyorchidism in children and their management plan. More specifically from Pediatric Endocrinology perspective, we discuss the influence of the Polyorchidism on the Tanner staging (by testicular volume), growth and pubertal development in boys.

Reference:
1. Amanda Myers, Bradley Morganstern and Ronnie Fine Urology, 2017-06-01, Volume 104, Pages 196-197,
2. Ibrahim H., Roberts M.J., and Hussey D.: Quadruple orchidopexy for torsion testis in an adolescent with polyorchidism: a case report. Urology 2016; 87: pp. 196-199.

Diabetes Mellitus and Glucose Metabolism

DIABETES COMPLICATIONS I

Involvement of NF-κB-p65 in BAG3 Regulation After Stress Stimuli

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SAT-632

Involvement of NF-κB-p65 in BAG3 Regulation After Stress Stimuli

We previously identified the limb salvage QTL1 (LSQ-1) on mouse chromosome 7 as a locus that offered protection against ischemic injury following induction of hind limb ischemia (HLI) a model of experimental peripheral arterial disease (PAD) in mice. To better understand the role of the LSQ-1 locus in post ischemic adaptation we characterized several genes within this locus and identified a number of genes that were important in tissue adaptation to ischemia, including BCL2-associated athanogene 3 (BAG3). BAG3 is an anti-apoptotic protein that plays an important role in cell survival through the regulation of autophagy. BAG3 expression is induced in the gastrocnemius muscles of mice after hind limb ischemia but how ischemia regulates BAG3 expression is poorly understood. Additionally, the activation of NF-κB transcription factor is essential for cell survival after stress stimuli. We hypothesized that BAG3 upregulation following stress stimuli is regulated by NF-κB. We determined whether NF-κB is involved in BAG3-mediated survival of primary human skeletal muscle cells (HSMC) during ischemia and diabetic conditions. Within 6 hours of treatment, ischemia induced BAG3 mRNA (no ischemia vs ischemia: 1.0 ±0.09 vs 1.41±0.02; $p<0.01$) and protein expression (BAG3/total protein, no ischemia vs ischemia 1.0±0.01 vs 1.38±0.06; $p<0.01$). Knockdown of BAG3 expression by shRNA induced early cell damage in HMSC under ischemic conditions as measured by HMGB1 expression (HMGB1/total protein; control plasmid vs shRNA, 1.0±0.09 vs 1.71±0.04; $p<0.01$). Knockdown of p65 subunit of the NF-κB by shRNA significantly diminished BAG3 mRNA expression after ischemia (control plasmid vs shRNA: 2.11±0.18 vs 1.48±0.05; $p<0.05$). Moreover, treatment of HSMC with 750 uM palmitic acid (PA) and 100nM insulin for 3 days to mimic diabetic conditions significantly increased the expression of BAG3 mRNA (control vs PAL+Ins, 1.0±0.14 vs 2.27±0.08; $p<0.01$) and protein (BAG3/total protein, control vs PAL+Ins, 1.0±0.03 vs 1.39±0.11, $p<0.01$). Knocking down p65 attenuated these increase in BAG3 mRNA (PAL+Ins vs shRNA+PAL+Ins, 2.27±0.08 vs 1.56±0.02 $p<0.01$) and protein (BAG3/total protein; PAL+Ins vs shRNA+PAL+Ins, 1.39±0.11 vs 0.99±0.1; $p<0.05$) Thus, 1) BAG3 expression is important
in cell survival under ischemic conditions, and 2) NF-kB plays a key role in upregulating the expression of BAG3 under diabetic and ischemic conditions.