novo 1p13.2 deletion which was diagnosed by array-CGH analysis.

Clinical Case: A Greek boy, who was referred for evaluation of growth failure, was investigated. He was delivered at term by cesarean section, with normal birth weight (3.060 kg), birth length (51 cm) and head circumference (35.5 cm). There was no positive family history for short stature. Target height was on the 25th percentile. He presented with growth deceleration since the age of 12 months leading to stature lower than the 3rd percentile at the age of two years, while his weight was at the 3rd percentile. Macrocephaly was appreciated (53 cm, >95th percentile). Dysmorphic facial features resembling Noonan syndrome, such as frontal bossing, depressed nasal bridge and low-set ears were recognized. His motor development was normal. Laboratory examination revealed hypothyroidism and treatment with L-thyroxine was initiated. He had regular follow-ups at 6 months intervals. A mild delay in speech development and motor skills was appreciated. At the age of five years old, as his growth rate remained slow, growth hormone (GH) stimulation tests were performed. GH had a borderline normal peak of 9.9 ng/ml. IGF-1 levels were also within normal range (123.7 ng/ml). Magnetic Resonance Imaging (MRI) of the pituitary gland and the brain was normal. Array-CGH analysis detected a loss of approximately 800 Kb on chromosome 1p13.2; these alterations affect 8 genes in the OMIM database. The deleted segment was mapped at chr1: 115,186,092_115,977,647 region. The genomic coordinates are listed according to genomic build GRCh37/hg19. The genetic material analysis did not show the presence of a deficit in chromosomal region 1p13.2 (chr1: 115,186,092_115,977,647) in neither of the parents, indicating that this finding was created de novo.

Conclusion: We describe a patient with a Noonan like phenotype, due to a 1p13.2 deletion. In the international literature and databases of Database of Genomic Variants and Database of Chromosomal Imbalance and Phenotype in Humans Using Ensemble Resources there are no reports of normal individuals or patients with a similar finding in chromosomal region 1p13.2. Regular follow up of the patient is needed in order to better understand the evolution of the phenotype.

Pediatric Endocrinology

ADVANCES IN PEDIATRIC OBESITY AND CANCER

Bone Outcomes Following Sleeve Gastrectomy in Adolescents and Young Adults with Obesity Versus Non-Surgical Controls

Madhusmita Misra, MD, MPH[1], Vibha Singhal, MD[2], Brian Carmine, MD[3], Amita Bose, BS[1], Megan Moriarty Kelsey, MD[4], Fatima Cody Stanford, MD[1], Jennifer Bram, MD[5], Jeremy Aidlen, MD[5], Thomas Inge, MD[6], Mary L. Bouxsein, PhD[7], Miriam Bredella, MD[1].

[1]Massachusetts General Hospital and Harvard Medical School, Boston, MA, USA, [2]Massachusetts General Hospital, Boston, MA, USA, [3]Boston Medical Center, Boston, MA, USA, [4]Children's Hosp Colorado/Univ of Colorado School of Medicine, Aurora, CO, USA, [5]University of Massachusetts Medical School, Boston, MA, USA, [6]Children's Hospital Colorado, University of Colorado Denver, Aurora, CO, USA, [7]Beth Israel Deaconess Medical Center, Boston, MA, USA.

OR22-06

Background: Sleeve gastrectomy is the most commonly performed weight loss surgery in adolescents with moderate-to-severe obesity. While studies in adults have reported on the deleterious effects of gastric bypass surgery on bone structure and strength estimates, data are lacking for the impact of sleeve gastrectomy on these measures in adolescents.

Objective: To evaluate the impact of sleeve gastrectomy on bone outcomes in adolescents and young adults over 12 months using dual energy x-ray absorptiometry (DXA) and high resolution peripheral quantitative computed tomography (HRpQCT).

Participants and Methods: We enrolled 33 youth 14-22 years old with moderate to severe obesity; 17 underwent sleeve gastrectomy and 16 were followed without surgery. DXA was used to assess areal bone mineral density (aBMD). HRpQCT was used to assess bone geometry, microarchitecture and volumetric BMD (vBMD) and finite element analysis to assess strength estimates (stiffness and failure load) at the distal tibia and distal radius at baseline and 12 months. 25(OH) vitamin D (25OHD) levels were obtained at baseline and follow-up.

Results: The surgical group lost 28.2% of total body weight compared to 1.4% in the non-surgical group. The groups did not differ for changes in 25OHD levels (P=0.181). After controlling for age and sex, compared to the non-surgical group, the surgical group had reductions in aBMD Z-scores at the femoral neck and total hip ($p \leq 0.0005$). At the distal tibia, there were reductions in cortical thickness and trabecular number, and increases in trabecular separation and cortical vBMD, without changes in strength estimates in the surgical group vs. controls ($p \leq 0.043$). Changes were less marked at the distal radius. While sleeve gastrectomy resulted in deleterious effects on most bone parameters, there was an increase in cortical vBMD at both sites, possibly from a decrease in cortical porosity. Most differences were attenuated after adjusting for changes in BMI over 12 months.

Conclusions: Over 12 months, weight loss associated with sleeve gastrectomy in adolescents had deleterious effects on areal BMD, bone geometry and trabecular microarchitecture at weight-bearing sites. However, strength estimates did not decrease, possibly because of a simultaneous increase in cortical volumetric BMD. Additional research is necessary to determine the relative contribution(s) of weight loss and the metabolic effects of surgery, and whether the observed effects on bone stabilize or progress over time.

Pediatric Endocrinology

PEDIATRIC SEXUAL DIFFERENTIATION, PUBERTY, AND BONE BIOLOGY

Support for a New Therapeutic Approach of Using a Low-Dose FGFR Tyrosine Kinase Inhibitor (Infigratinib) for Achondroplasia

Benoit Demuynck, Engineer[1], Justine Filipo, Engineer assistant[1], Gary Li, PhD[2], Carl L. Dambkowski, MD, MA[2], Laurence Legeai-Mallet, PhD[1].

[1]INSERM U1163, Imagine Institute, Paris University, Paris, France, [2]QED Therapeutics, San Francisco, CA, USA.