SLEP SUMMER SCHOOL 2020

Fellow 7 Maria Paula Sarmiento

A 8 7/12 years old male patient without any relevant family history. He was born via uncomplicated vaginal delivery to a G1P0 mother. At 18 months of age was diagnosed with primary hypothyroidism in the setting of generalized hypotonia and failure to meet his neuro-developmental milestones. Additional findings at that time included microcephaly, short stature, genu valgus, joint hypermobility and a marfanoid habitus. He has been on levothyroxine supplementation since diagnosis.

Initial genetics studies showed an karyotype 46,XY and the Allgrove syndrome and mucopolysaccharidosis were ruled out. At 8 years old, an exoma trio confirmed a *de novo* M918T heterozygous pathogenic variant in the *RET* gene. Lab tests was ordered. A CT of the chest and abdomen was performed which showed a right paratracheal cervico-thoracic mass with a 30 x 35 x 36 mm upper mediastinum component.