

Table 1. Clinical and metabolic parameters of the patient carrying in heterozygosity the novel MC4R p.Met215del and the known FTO rs9939609 SNP

Age (y)	Height (SDS)	BMI (SDS)	BA (y)	*BP (mmHg)	Kcal/day	**Total Cholesterol/ HDL/LDL/TRG (mg/dl)	Glucose (mg/dl) (70-99)	Insulin (IU/l) (<15)	HOMA <2	Other hormones
0.6	+2.5	+1.8				153/46/89/101 (50 th centile)				
3	+3.6	+4.5	5	75/45 (75 th centile)	2500	183/54/99/148 (90 th centile)	78	10.5	1.8	Free T4:1.55 ng/dl (0.79-1.76) TSH: 2.77 mIU/l (0.4-5.5) Cortisol am: 8.16 micg/dl (5-25) ACTH am: 11.4 pg/ml (7.2-6.3) Prolactin: 20.6ng/ml (5-29)
5.2	+4.6	+3.1	8.5	110/70 (97 th centile)	3100	200/58/123/150 (95 th centile)	76	14.6	2.4	25 (OH) D3: 12ng/ml (insuf>30) IGF1: +2SDS, IGFBP3: +2SDS OGTT (Min): 0, 30, 60, 90, 120 Glucose: 76, 110, 122, 95, 89 Insulin: 14, 170, 154, 123, 110
6	+4.3	+2.8		90/50 (50 th centile)	2500	180/60/110/130 (90 th centile)	88	11.5	2.3	Free T4: 1.32 ng/dl (0.79-1.76) TSH: 3.14 mIU/l (0.4-5.5) Cortisol am:11.16 micg/dl (5-25) ACTH am:15.4pg/ml (7.2-6.3)

*Percentiles for blood pressure from: The fourth report on the diagnosis, evaluation, and treatment of high blood pressure in children and adolescents. Pediatrics 2004;114 (2):555-76.

** Percentiles for lipid profile from: Lipids screening and cardiovascular health in childhood. Pediatrics. 2008;122(1):201.