CLINICAL CASE 3

CLINICAL HISTORY

45 years old woman, with android obesity type, centripetal.

She reports that she doesen't practice physical exercice, eats junk food, smokes, drinks one beer and one glass of wine daily.

She was a girl with normal weight, at adolescence she put on 10 kg and after her two pregnancies she gain 10 kg more.

Feels fluid retention and pain in legs, notices that she has no waist anymore.

She feels lack of attention and slow to understand her children's homework.

She has never been to the doctor and decides to visit a specialist to lose weight.

RECENT HISTORY:

She currently is not feeling well.

She feels discouraged, works hard and has no time to eat at lunch time, arrives home very anxious, grabs a bite of pastries and anything she finds at hand, prefers something sugary than salty.

Weight: 88 kg

Height: 158 cm

BMI: 35,3

Impedanciometry: Fat: 36% (31,7 Kg).

Blood pressure: 15/9

GENETIC STUDY

Performed by DNA extraction from oral mucosa sample on swab, followed by DNA amplification by PCR and DNA analysis with molecular biology techniques. 21 genes were studied.

Results of the genetic study are presented in table 1.

GENE	VARIANT ANALISED	PATIENTS'S GENOTYPE	ASSOCIATED WITH:
MC4R	rs17782313T>C	TT	
MTHFR	c.1286A>C (1298A>C)	AA	
FTO	c.46-23525T>A	TT	
MTHFR	c.665C>T (677C>T)	CT	Decreased activity of methylenetetrahydrofolate reductase enzyme.
FABP2	c.163A>G (p.T55A)	GA	Insulin resistance and risk of atherosclerosis.
BDNF	c.196G>A (p.Val66Met)	GG	

Gene	Variant	Genotype	Description
UCP1	c.-3826A>G	GG	Together with the R64 variant in gene ADRB3, tendency to weight gain in adulthood.
ADIPOQ	c.214+62G>T	TT	Insulin resistance and risk of coronary heart disease.
APOE	*E3/*E4	E4/E4	Higher risk of hypercholesterolemia and ischemic heart disease.
UCP3	c.-2078C>T	CC	
ADRB2	c.79G>C (p.Q27E)	CC	
PAI1	c.-675_4G/5G	4G/5G	Promotion of the formation of atherosclerotic plaques and reduced fibrinolysis, responsible for acute myocardial infarction.
ADRB3	c.190T>C (p.W64R)	TT	
IRS1	c.2911G>A (p.Gly971Arg)	GG	
PPARG	c.34C>G (p.P12A)	CC	Ancestral genotype not adapted to current eating habits. Increased risk of insulin resistance, diabetes and obesity.
NOS3	c.894T>G (p.Asp298Glu)	TG	Higher incidence of essential hypertension and independent increased risk of coronary artery disease (CAD).
ACE	IVS16-407ins(289nt)	ID	Increased body weight and blood pressure.
IL1B	c.315C>T	CC	Increased total fat mass. Altered immune function of IL-1 and obesity development.
IL1RN	IVS4-515ins(86nt)	*4	
IL6	c.116-121C>G	CG	
GNB3	c.825C>T	CT	Higher Body Mass Index (BMI) and increased risk of progression towards more severe hypertension.
TNFa	c.-175-313G>A	GG	

Table 1. Patient's genetic results

RESULT

Result: very high genetic load

According to the results obtained in this analysis, we can conclude the following:
- Good food intake control and some protection against stress.
- Acceptable thermogenesis control and good basal metabolism.
- Moderate-low inflammatory profile.

Risk of insuline resistance and Type II Diabetes, moderate to high.

Regarding the group of Cardiometabolic Risk Genes (GNB3, MTHFR (2), APOE, PAI1, NOS3) the patient presents 5 of the 6 thrifty polymorphisms, 1 in homozygosis (APOE) and 4 in heterozygosis (GNB3, MTHFR(2), PAI1, NOS3). The patient's cardiometabolic risk is moderate to high.

Regarding the group of Arterial Hypertension Risk Genes (ADRB2, ADBR3, GNB3, ACE) the patient presents 2 of 4 thrifty polymorphisms, of which 2 in heterozygosis (GNB3, ACE). The patient has a moderate risk of hypertension.

Regarding the group of Senile dementia and Alzheimer's Disease Risk Genes (FTO, APOE, PAI1, NOS3, IL6, ACE) the patients presents 5 of 6 polimorfismos altered, 3 of them in heterozygosis (PAI1, NOS3, IL6), 1 in homozygosis (APOE 4) and ACE (ID) in synergy with APOE 4. The patient presents a high risk.

It is important to start early treatment with atorvastatin and hypolipemiants, in order to neutralize the effect of APOE 4.

EVOLUTION

It is too soon to assess the importance of early intervention in these type of patients, the important thing is that thanks to genetic testing for obesity control, we have discovered that what matters is the cardiometabolic control.

It was difficult to convey the information to the patient, without frightening her but making her understand the importance of following rigorously the medication and diet instructions.

Today, she has lost weight and her blood pressure levels are normal.