CASE 023

CŌRE DIAGNOSTICS™

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Monogenic Obesity in the World's Heaviest Woman: A Case Report.

CASE HISTORY

A 36 year old female of Arab-Egyptian ethnicity presented with symptoms of Metabolic Syndrome. She weighed 504 Kgs, and had history of smoking, cerebrovascular accident (CVA), chronic kidney disease (CKD), bronchial asthma (BA), chronic obstructive pulmonary disease (COPD), and hypothyroidism. She had no history of diabetes or cancer. The patient had early onset obesity, with exponential weight gain despite dietary modification. The patient was on Eltroxin treatment at the time of testing, and was scheduled for bariatric surgery.

INVESTIGATION

The patient was tested in a 36 gene NGS panel for both autosomal recessive and autosomal dominant forms of monogenic obesity. The results were confirmed via Sanger sequencing.

METHODOLOGY

The obèCORE monogenic obesity panel by Next Generation Sequencing [NGS]: This test comprises sequence analysis of genes associated with monogenic obesity: *ALMS1, ARL6, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, CEP290, CRTC1, CUL4B, DYRK1B, GNAS, LEP, LEPR, MAGEL2, MC3R, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, POMC, PPARG, PYY, SDCCAG8, SIM1, TRIM32, TTC8, UCP3, VPS13B*