CASE 031

CŌRE DIAGNOSTICS™

MALE BREAST CANCER: A CASE REPORT WITH A GERMLINE MUTATION IN *BRCA2* GENE

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PAST HISTORY: A 55-year-old man was diagnosed with hormone-receptor-positive breast cancer in 2013. He presented with diabetes mellitus, hypertension, and hypercholesterolemia as comorbidities. At the time of testing, he was on Tamoxifen standard adjuvant therapy.

TEST FINDINGS

We carried out gene sequencing BY NGS/MPS, and MLPA analysis [deletion/duplication analysis] for BRCA1:BRCA1, DNA repair associated, and BRCA2:BRCA2, DNA repair associated genes in the patient. The patient tested positive for a heterozygous mutation in *BRCA 2* gene, namely: NM_000059.3(BRCA2):c.6634_6637delTGTT (p.Cys2212Leufs). The cytogenetic location of this abnormality is: 13q13.1, and the genomic location: Chr13: 32340989 - 32340992 (on Assembly GRCh38). Its molecular consequence is a frameshift_variant [A sequence variant which causes a disruption of the translational reading frame, because the number of nucleotides inserted or deleted is not a multiple of three]. It is classified as pathogenic (class 1) as per ACMG recommendations. ClinVar (Accession: RCV000241285.3) lists this variant as pathogenic based on 3 submissions: SCV000301079, SCV000327480, and SCV000733286.