**Variants and genes associated with carrier health risk:**

**The importance of carrier screening beyond family planning**

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**Objective:** Carrier screening panels typically do not include any autosomal dominant (AD) disorders, although some heterozygous carriers of certain autosomal recessive (AR) disorders may manifest symptoms because in some cases, genes could lead to an AD condition in the same gene. In addition, there are patients who may have variants in compound heterozygous or homozygous for AR conditions, leading to an additional genetic counselling dilemma. This study aims to assess the percentage of cases with variants in genes that cause genetic conditions of AR or X-linked (LX) inheritance that could lead to health risks for carriers. **Methods:** Retrospective study of carrier genetic tests (Horizon 4 or 274 genes) performed for family planning before assisted reproduction treatment at DASA GENÔMICA between 2021 and 2022 (24 months). We evaluated the results in two scenarios: (1) cases that presented two variants in the homozygous or heterozygous compound state in all the genes studied; (2) whether patients with a positive test result had any pathogenic or probably pathogenic variant in the following genes reported in the literature and/or databases associated with health risk: ATM (increased risk of cancer, especially breast); NBN (increased risk of cancer, especially breast); GBA (Parkinson's disease); COL4A4 (hematuria); GJB2 (deafness with AD inheritance); CFTR (pancreatitis); FMR1 (ovarian insufficiency); DMD (cardiomyopathy); GLA (heart disease and hypertension). The methodology used in the panels was next-generation sequencing (NGS) and genes such as FMR1 and DMD were evaluated by complementary techniques. The study consisted of 167 patients (77.84% women and 22.16% men), aged 18 years or older, with no report or knowledge of genetic diseases in the family. **Results:** 68 patients had at least one detected pathogenic or probably pathogenic variant of all analyzed genes (4 or 274 genes, depending on the panel). In three patients, two variants were detected in different genes: GJB2 related to deafness; CFTR related to cystic fibrosis, and GAA which could lead to Pompe’s disease. However, it was not possible to determine whether they were in cis (same allele) or trans (different allele). We observed 5 patients with a variant in GJB2 (sensorineural deafness); 4 with variants in CFTR; 2 patients with GBA variant and 1 patient with 70 CGG repeats in FMR1. **Conclusions:** This study demonstrates that approximately 8% of the patients presented variants in genes that may increase the health risk for the individual and not just for the offspring, although carrier screening panels do not aim to detect or diagnose conditions in patients. Such disorders deserve special attention and direct genetic counselling not only for family planning but also for monitoring the individual to stratify and reduce the risk of developing genetic alterations throughout life.