Propositions

belonging to this thesis

Improving the risk assessment of inherited breast and ovarian cancer: clinical significance of BRCA1/2 variants and risk modifiers

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1. Populations with whom the Portuguese have mixed such as those from the Portuguese-speaking African countries should be screened for the Portuguese founder mutation *BRCA2* c.156_157insAlu. (this thesis)

2. The opposite effects of *FGFR2* rs2981582 polymorphism in breast and ovarian cancer can be explained by the fact that FGFR2 is a progesterone receptor activator. (this thesis)

3. The pathogenic effect of rare sequence variants on splicing, beyond those on the ag/G and AG/gt consensus sequences, is underestimated. (this thesis)

4. A genetic classifier based on biological processes is more reproducible in different datasets than classifiers based on the most statistically significant differentially expressed genes. (this thesis)

5. For *BRCA1/2*-mutation carriers, personalized medicine is not only about determining the best risk-reducing options, but also about their best timing.

6. It is important to comprehend all the naturally-occurring BRCA1/2 transcript isoforms, since the normal must be known to allow to recognize the abnormal.

7. The potential of next-generation sequencing of RNA (RNA-seq) surpasses that of the DNA-seq.

8. In the future we will carry around our own whole-genome sequence in our cell phone.

9. Family and health often go together.

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