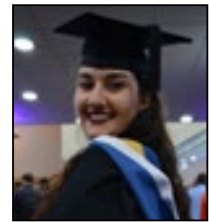


Creating a MySQL database for mutations and variants associated with breast cancer for the investigation of its link to primary infertility



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Biography

Ilektra Vasiloudi has successfully completed the MSc in Prenatal Genetics and Fetal Medicine at University College London. During her studies she focused on topics like (IVF), gametogenesis, fetal medicine and genetics, prenatal diagnosis and screening as well as preimplantation genetic testing. At the same time, she attended annual conferences and events on embryology and fertility in London and was involved at a conference in Edinburgh, Scotland celebrating the 40 years of IVF. Miss Vasiloudi has been trained on genomic counseling skills as she wants to follow this career path. Her latest research focused on breast and ovarian cancer and its link to primary infertility. She is passionate about the wellbeing of women and keeps supporting charities and fundraisers that revolve around women's and children's health.

Background

For women of reproductive age, a new diagnosis or an increased risk of breast cancer, can lead to important changes in fertility. At the same time, women who have infertility issues usually follow the in vitro fertilization (IVF) pathway, where fertility drugs are used to stimulate hyperovulation. Although, studies that have been done in measuring long-term risk of breast cancer after IVF have produced unclear results (mainly due to methodological limitations), hormonal exposures raise a serious concern for breast cancer risk. Most studies do not present a significant risk after the use of fertility medications, but there are still some other publications that pose a possible elevated risk.

Aim

The main aim of this project was to test the hypothesis that there was a correlation between variants of breast cancer and infertility genes. In order to perform that, the aims were to identify the genes of interest and the available variants as well as built a database using MySQL and other bioinformatic tools.

Results

The total amount of data collected for human variants for breast cancer was 9288 linked with 11 genes (including TP53, CDH1, PTEN) and 1340 for infertility associated with 13 genes. After no link was found at the gene and variant levels of research, signaling pathway level was attempted, but still there was no clear correlation between breast cancer and infertility. On a positive note, interesting findings were discovered around clinical significance of variants and BRCA gene polymorphisms.

Conclusion

In order to simplify the complexity that this topic presents, more studies on more populations are needed to clarify and address the controversial results. The common signaling pathways identified surely spark new possibilities for research and a more thorough investigation could lead to novel findings, even a breakthrough. Only then, a general reassuring idea will be available for both scientists and clinicians as well as for the affected populations.



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