

Abstract Title: Building a Human Infertility Genome Database

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Introduction: Infertility is a multifactorial clinical phenotype that approximately affects 10%-15% of the global population and still poses as a common problem for many couples. The work into genomics and infertility is still ongoing to date showing how complexed infertility is in both idiopathic and genomic related diseases. We created and currently analysing a novel SQL database consisting of 50 patients with fertility complications. Preliminary data we provide are cumulative genome annotation data presented as four major datatypes: CNVs, SVs, InDels, and SNPs.

Aim: To identify the chromosome that occurs frequently amongst our datasets within the database for future analysis.

Methods: We categorised the patients into categories: recurrent miscarriage, recurrent implantation failure, advanced maternal age, azoospermia, oligoasthenoteratozoospermia, asthenospermia, poor sperm parameters, and normal sperm parameters that required ICSI. We then identified the most common chromosomal locations with flagged variants amongst the groups. We queried the data to gain an accumulated chromosome count per chromosome for each of the categories and normalised the data.

Results: From our data queries and data normalisation, various chromosomal regions have been flagged up, with chromosome 19 regions most frequently. A review of the literature revealed the need for more research into these chromosomal areas and their relationship to infertility.

Conclusion: Amongst the data within our database, we identified various variants frequently flagged on specific chromosomal regions. Currently, we are continuing working in our approach to uncover genomic locations that relate to human infertility and link those to other genomic studies and databases.