P-024 Identification of spermatogenic infertility phenotypes using next generation sequencing

Study question:

- Wider implications of the findings: mutants associated to spermatogenic failure are needed. Advances in NGS allowed us to study a large number of genes involved in spermatogenesis process in patients with idiopathic infertility.

- Summary answer: Assessment of semen parameters is very important. The following indicators were evaluated: amplitude lateral head displacement (ALH), best cross frequency (BCF), VAP/VCL. There were significant correlations between normal fertilization rates and sperm parameters: 60% normal fertilization (group 1: 60 cycles; group 2: 71 cycles). Sperm motility before insemination was more measured by SMAS as an indicator of normal fertilization rates. However, there were no significant differences in most baseline characteristics across groups, except for the criterion of sperm motility characteristics assessed by SMAS. In this study, we investigated the utility of the movement characteristics of motile sperm assessed by SMAS are related to the functional capacity (ICSI). Several studies have reported that objectively movement characteristics of motile sperm before insemination is useful as an indicator in determining what is known already: normal fertilization rates after in vitro fertilization (IVF).

- Main results and the role of chance: The main limitation of this study is the retrospective design and small sample size. A prospective study is necessary for the selection of methods for fertilization in vitro.

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- Limitations, reasons for caution: Wider implications of the findings: mutants associated to spermatogenic failure are needed. Advances in NGS allowed us to study a large number of genes involved in spermatogenesis process in patients with idiopathic infertility.

- Trial registration number: P-024

- Participations/materials, setting, methods: A retrospective study was performed from April 2018 until July 2020. A total of 30 patients with abnormal seminal count, 20 healthy donors selected on the basis of normal semen parameters, and 10 patients carrying Y-chromosome microdeletions or abnormal karyotype were excluded. The control group included 20 normal men with normal count and morphology of sperm. Patients carrying Y-chromosome microdeletions, autosomal deletions, and chromosome Y microdeletions. However, current genetic studies explain only 4% of cases, whilst most cases of male factor infertility remain without a clear diagnosis. Therefore, new techniques that explain the cause of male infertility are needed. Advances in NGS allowed us to study a large number of genes involved in spermatogenesis process in patients with idiopathic infertility. Next generation sequencing (NGS) has become a method to identify new genetic causes of male infertility for the development of new therapies. In the present study, patients were selected on the basis of normal semen parameters and 38th Hybrid Annual Meeting of the ESHRE, Milan — Italy, 3–6 July 2022

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Study question: To evaluate the efficacy of the method of freezing the seminal sample at home by the patient himself.

Summary answer: The freezing of the seminal sample at home is an effective method that brings the clinic closer to the patient, eliminating travel and stress.

What is known already: Seminal sample freezing for reproductive purpose is indicated in different situations: preservation of fertility, before a vasectomy, in patients with dysfunctions and problems obtaining a sample, and for those patients who cannot be present at the time of the oocyte retrieval.

In many of these cases, the patient is under pressure to obtain the sample, in a clinical environment and at an exact time. Many experience this as a stressful situation, which in some cases forces them to travel if they live far from the clinic.

Study design, size, duration: In 2017, the home freezing protocol was validated with volunteers who obtained a semen sample at the clinic. After verifying that the quality of the sample was the same as if it were frozen by an expert biologist, we began offering this technique to our patients in 2019. We compared the results of the DO cycles with a control group to determine the efficacy of the protocol.

Participants/materials, setting, methods: Until December 2021, a total of 18 patients have used the Freezing Kit at home, 14 were residents in other EU countries, and 4 residents in Spain, but far from the clinic. As a control group, we used 608 oocyte donation cycles with a frozen sample.

Patients who decided to freeze their sample at home receive a kit consisting of a travel tank with nitrogen vapors, a box with all the necessary material, and instructions.

Main results and the role of chance: Regarding the results of the 18 patients who have used the kit to freeze samples for their oocyte donation cycles, they have a mean of 44 ± 7.1 years, and the samples had a post-thaw concentration of 48 ± 30.5 M/ml with a mean mobility of 21.18 ± 14.6%.

No statistically significant differences were found in the fertilization rate compared to the control group (76.4 ± 15.1% vs 77.8 ± 18.3%; p = 0.485), blastocyst rate (61% ± 10.5% vs 56.1 ± 16.4%; p = 0.387) nor clinical pregnancy rate (62.8% vs 47.6%; p = 0.251).

The fact of having a live birth from a sample frozen by the patient himself has just demonstrated the safety and usefulness of this novel freezing protocol.

Limitations, reasons for caution: The number of patients who have undergone this process is limited due to the nature of the treatment.

Wider implications of the findings: These results demonstrate the clinical utility of this freezing method for certain patients, where travel to the clinic may be inconvenient. Demonstrating that it is an effective method of cryopreservation of the seminal sample that brings the clinic closer to the patient, eliminating trips and stress.

Trial registration number: not applicable