The why, the how and the when of PGS 2.0: current practices and expert opinions of fertility specialists, molecular biologists, and embryologists

Karen Sermon Antonio Capalbo Jacques Cohen Edith Coonen Martine De Rycke Anick De Vos Joy Delhanty Francesco Fiorentino Norbert Gleicher Georg Griesinger ... Show more

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Abstract

STUDY QUESTION

We wanted to probe the opinions and current practices on preimplantation genetic screening (PGS), and more specifically on PGS in its newest form: PGS 2.0?

STUDY FINDING

Consensus is lacking on which patient groups, if any at all, can benefit from PGS 2.0 and, a fortiori, whether all IVF patients should be offered PGS.

WHAT IS KNOWN ALREADY

It is clear from all experts that PGS 2.0 can be defined as biopsy at the blastocyst stage followed by comprehensive chromosome screening and possibly combined with vitrification. Most agree that mosaicism is less of an issue at the blastocyst stage than at the cleavage stage but whether mosaicism is no issue at all at the blastocyst stage is currently called into question.

STUDY DESIGN, SAMPLES/MATERIALS, METHODS

A questionnaire was developed on the three major aspects of PGS 2.0: the Why, with general questions such as PGS 2.0 indications; the How, specifically on genetic analysis methods; the When, on the ideal method and timing of embryo biopsy. Thirty-five colleagues have been selected to address these questions on the basis of their experience with PGS, and demonstrated by peer-reviewed publications, presentations at meetings and participation in the discussion. The first group of experts who were asked about 'The Why' comprised fertility experts, the second group of molecular biologists were asked about 'The How' and the third group of embryologists were asked about 'The When'. Furthermore, the geographical distribution of the experts has been taken into account. Thirty have filled in the questionnaire as well as actively participated in the redaction of the current paper.

MAIN RESULTS AND THE ROLE OF CHANCE

The 30 participants were from Europe (Belgium, Germany, Greece, Italy, Netherlands, Spain, UK) and the USA. Array comparative genome hybridization is the most widely used method amongst the participants, but it is slowly being replaced by massive parallel sequencing. Most participants offering PGS 2.0 to their patients prefer blastocyst biopsy. The high efficiency of vitrification of blastocysts has added a layer of complexity to the discussion, and it is not clear whether PGS in combination with vitrification, PGS alone, or vitrification alone, followed by serial thawing and eSET will be the favoured approach. The opinions range from in favour of the introduction of PGS 2.0 for all IVF patients, over the proposal to use PGS as a tool to

rank embryos according to their implantation potential, to scepticism towards PGS pending a positive outcome of robust, reliable and large-scale RCTs in distinct patient groups.

LIMITATIONS, REASONS FOR CAUTION

Care was taken to obtain a wide spectrum of views from carefully chosen experts. However, not all invited experts agreed to participate, which explains a lack of geographical coverage in some areas, for example China. This paper is a collation of current practices and opinions, and it was outside the scope of this study to bring a scientific, once-and-for-all solution to the ongoing debate.

WIDER IMPLICATIONS OF THE FINDINGS

This paper is unique in that it brings together opinions on PGS 2.0 from all different perspectives and gives an overview of currently applied technologies as well as potential future developments. It will be a useful reference for fertility specialists with an expertise outside reproductive genetics.

LARGE SCALE DATA

none.

STUDY FUNDING AND COMPETING INTEREST(S)

No specific funding was obtained to conduct this questionnaire.

preimplantation genetic screening, blastocyst biopsy, array comparative genomic hybridization, massive parallel sequencing, vitrification, chromosomal abnormalities, preimplantation embryo

Topic: biopsy fertilization in vitro belgium embryo stage 3 structure china chromosomes embryo fertility germany greece italy netherlands peer review reproductive physiological process social role spain technology genetics mosaicism genetic analysis comparative genomic hybridization genetic screening biopsy of embryo cytokinesis funding transfer technique consensus setdb1 gene

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