PGDIS Newsletter, 13 September, 2018

18TH INTERNATIONAL CONFERENCE ON PREIMPLANTATION GENETICS, Geneva, Switzerland, April 15-18, 2019

PROVISIONAL SCIENTIFIC PROGRAM

Dear Colleague,

The review of the extended application of NGS based PGT-A provided an increasing body of data on sub-chromosomal copy number variations and mosaicism in relation to their clinical impact, which will form the basis for updating PGDIS recommendations on the topic. As there is still insufficient understanding of their biological and clinical significance, it will be further explored at the 18th International Conference on Preimplantation Genetics, to be held in Geneva, Switzerland, April 15-18, 2019 (see Scientific Program on PGDIS website). While higher resolution PGT-A is still required for more accurate detection of these variations, the accumulated data will be further explored with the aim of clarifying the interpretation of these abnormalities and provide a basis for genetic counseling of PGT-A patients.

The origin and mechanisms of mosaicism and segmental aneuploidies will be further explored to allow a better prediction of chances of the embryos to implant and produce a viable pregnancy. Also, the chromosomal status of the embryo may not be the only selection criterion to improve pregnancy outcomes. So the additional tests to improve selection of euploid embryos will be addressed. Among the candidate tests being explored are mitochondrial DNA, epigenetic and genetic expression profiles, as well as time-lapse imaging and endometrial receptivity, the utility of which is still not fully understood and even controversial, requiring a further critical evaluation.

Among other emerging technologies to be explored for the first time will be preconception carrier screening and recent developments in whole genome sequencing and their impact on the application of PGTin clinical practice. The challenge of whole genome sequencing, particularly at the single cell level, will be to identify pathological copy number variants and other mutations of previously unknown significance and highlight the need for improving the approaches for PGT of *de novo* mutations.

Although the law regarding PGT in Switzerland, the venue of the Conference next year, is in progress, it remains restricted in Switzerland, Germany and Austria. While the ongoing revision will allow genetic testing of embryos at risk for a severe genetic disorder, the topic will be addressed in a special session devoted to preconception polar body-based PGT for both

monogenic disorders and aneuploidy testing. Evidence of a possible detrimental effect of invasive biopsy procedures has resulted in increasing interest in the development of non-invasive PGT (NIPGT), particularly for PGT-A, which will be explored in more detail. Also, with the recent success of non-invasive prenatal testing (NIPT), as a follow up confirmatory test for PGT, the accumulated results will be analyzed for the reliability and accuracy of the test, to explore the prospect of replacing follow up invasive prenatal diagnosis, which is still recommended after PGT.

Finally, with the progress in single cell DNA methylome sequencing of human preimplantation embryos, feasibility of PGT for epigenetic disorders will be addressed, as well as the advances in the related areas of research, such as in CRISPR-based gene editing for their potential utility as an alternative to PGT for monogenic diseases.

As usual a number of Pre-congress Workshops will be organized, including three Pre-Congress courses devoted to the main trends in PGT technological developments, as well as the Annual Hands-On Workshop on PGT by Blastocyst Biopsy, April 11-13, 2018 at the Istanbul Memorial Hospital PGT Center (see Hands-On Workshop program on the PGDIS website).

The progress in the organization of the Conference may be followed on the PGDIS website (www.pgdis.org), with the registration and abstract submission to be opened January, 2019.

We are looking forward to welcoming you in Geneva!

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