PRE-CONGRESS WORKSHOP
GENETIC COUNSELING IN PREIMPLANTATION GENETIC DIAGNOSIS (PGD)
Miami Beach, Florida, April 23, 2009 (13:00-17:00)

Chair: Joe Leigh Simpson, Florida International University, Miami, FL

Presenters: Christina Levin, Reproductive Genetics Institute, Chicago, IL
Jill Fischer, Reprogenetics, West Orange, NJ
Rene Genovese, Reproductive Genetics Institute, Chicago, IL
Dana Pauling, Reproductive Genetics Institute, Chicago, IL

Workshop Outline:

With the application of PGD to genetic practices and reproductive medicine, genetic counseling is becoming the key component, not only due to the novelty of the technology, but also because of the need to develop the custom made design of PGD for each couple. Also there is an expanding need for providing sophisticated information of specific risk figures and error rates involved, depending on the status of the technology, which is still under extensive development. There are also a few PGD indications that have never been used in prenatal diagnosis, such as the late onset disorders with genetic predisposition and preimplantation HLA typing. This requires additional explanation of the practical relevance of detection and transfer of mutation free or HLA matched embryos, because some predisposing genes carriers may never develop a disease, while pre-selection of HLA matched embryos benefit only the affected sibling in the family, requiring stem cell transplantation. Finally, with an increasing role of PGD as source of human embryonic stem cell (hESC) lines with genetic disorders, it is becoming useful to provide the at risk couples with the option to donate their affected embryos for derivation of hESC lines, required for discovering the primary defect needed for the development of the treatment regiments for genetic disorders. So the increasing role of genetic counseling in PGD will be demonstrated for the first time in more detail, to improve awareness of PGD impact on medical practice, which will contribute in the development and application of the appropriate genetic counseling services within the genetic practices and assisted reproductive technology.

Learning Objectives:

At the conclusion of this Workshop, the participants should be able to:

(1) Evaluate the importance of genetic counseling for PGD and its impact on PGD for different indications.

(2) Outline limitations of PGD and their practical relevance in performing genetic counseling in couples at risk for producing the progeny with Mendelian conditions, translocation and aneuploidy.

Provisional Program:

1. Why PGD versus PND
    a. Historical overview of PND and PGD
        i. Amnio
        ii. CVS
        iii. PGD by bb
        iv. PGD by PBR

v. PGD by blastocyst bx
b. “ethically more acceptable”
c. Timing of testing; pre-conception
d. Differentiation of risks re: dx (embryo loss vs pregnancy loss)

2. Techniques utilized in PGD
    a. Review of ART timing/technologies
        i. PBR
        ii. BB
        iii. Blastocyst biopsies
    b. PGD assays
        i. FISH
        ii. PCR
        iii. CGH

3. Indications for PGD
    a. Aneuploidy
        i. AMA
        ii. PCA
        iii. Failed IVF
        iv. Gender selection/family balancing
    b. Single Gene Disorders
        i. Non-traditional (late-onset/predisposition)
        ii. HLA
    c. Translocations
        i. Robertsonian
        ii. Reciprocal
        iii. Use of conversion technology

4. Genetic Counseling for the PGD patient
    a. Complex psycho-social needs
    b. Lack of understanding of ART
    c. Preliminary work-up and consent process
    d. Discussion of PGD and its risks/benefits/limitations
        i. Known risks of ART (multiple gest, etc)
        ii. Unknown risks of ART and/or PGD
        iii. Accuracy, need to consider PND
    e. Cost/insurance issues
    f. Unrealistic expectations re: success
    g. GC as coordinator of PGD
        i. Patient consultation
        ii. Laboratory set-up
        iii. Coordination of cycle, including work-up
        iv. Interpretation of and relaying results
        v. Follow-up (pre- and post-natal)
        vi. Future options

5. Panel discussion/Interesting case presentations