Reproductive genetics at the crossroads of the European Society of Human Reproduction and Embryology and the European Society of Human Genetics: an update

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Many questions regarding reproductive genetic issues are at the crossroads of both the European Society of Human Genetics (ESHG) and the European Society of Human Reproduction and Embryology (ESHRE). In 2005, a workshop was held in Seville to discuss these topics. This effort has resulted in the production of an impressive background document and the formulation of professional recommendations. ESHG published both the background document and the recommendations in the May 2006 issue of the European Journal of Human Genetics (Soini et al., 2006). ESHRE published the background document as a monograph and the recommendation paper was published in Human Reproduction (ESHG and ESHRE, 2006).

Since the developments in this area are very rapid, both societies felt it necessary to organize a follow-up discussion to the Seville workshop. This 2-day invitational meeting was held in Brussels on 5 and 6 March 2012. The 22 participants from both societies have produced a very comprehensive paper with the new developments in the field, which has been published in the European Journal of Human Genetics (Harper et al., 2013) and as a commentary paper in this issue of Human Reproduction and in full online (Harper et al., 2014).

Since the first report, the line of evidence that assisted reproductive technology (ART) is most probably associated with a slightly increased risk of imprinting disorders is growing, but the causes of these defects are not that clear and are difficult to be attributed. However, absolute risks appear to be low.

A meta-analysis of all published randomized controlled trials (RCTs) comparing IVF with and without preimplantation genetic screening (PGS) revealed that cleavage stage PGS using fluorescent in situ hybridization significantly lowered live-birth rates after IVF for women of advanced maternal age. Time and the results of RCTs will tell us if other biopsy and molecular approaches are of an advantage and, if so, for which indication groups.

Novel genetic and genomic technologies are likely to further change the testing of embryos and will not only result in generic methodologies to identify and select against embryos carrying disorders, but will indirectly result in a genome-wide view of the future of the developing embryo. Human embryonic stem cells and induced pluripotent stem cells that carry a particular mutation represent promising new disease models, especially for those diseases for which no good animal models exist.

In the near future the interface between ART and genetics will certainly become more important by the increase in our understanding about the genetics of infertility and the ability to perform more comprehensive genetic testing. At the moment there is not yet a routine ‘diagnostic’ indication for exome/whole genome analyses in male or female infertility, i.e. beyond the research setting. Furthermore, with respect to genetic testing in relation to reproduction, major legal differences exist in Europe. More and more patients find their own way out. Some do this by making use of cross border reproductive care, as has been shown by the ESHRE Taskforce, which confirmed that one of the main reasons for going abroad were legal restrictions. In other cases it is possible to opt for direct-to-consumer genetic testing which can be defined as the advertising and selling of genetic tests directly to consumers. These recent new developments also create various new challenges. Therefore it is to be expected that a third collaborative action of ESHG and ESHRE will be necessary in the not too distant future.

References

Editorial Commentary
