Commentaries

Designer babies – are they a reality yet?

Case report: simultaneous preimplantation genetic diagnosis for Fanconi anaemia and HLA typing for cord blood transplantation

Yury Verlinsky, Svetlana Rechitsky, William Schoolcraft, Charles Strom and Anver Kuliev
Reproductive Genetics Institute, Chicago, Illinois, USA

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HLA (human leukocyte antigen) matching for pre-selection of a potential donor progeny for transplantation has never been an indication for traditional prenatal diagnosis because pregnancy termination would likely be necessary. On the other hand, preimplantation HLA testing should be acceptable for this purpose because only HLA-matched pregnancies are established.

We performed the first such successful case, resulting in pre-selection and transfer of only those unaffected embryos that represent HLA match for sibling at need for HLA identical cord blood transplantation. The healthy unaffected child has been born, from whom umbilical cord blood was collected and transplanted into the 6-year old sibling with Fanconi anaemia. So this is really the first child in the world, born after preimplantation diagnosis for Fanconi's anaemia, who is also a perfect HLA match for the affected sibling, providing cord blood stem cells for transplantation. The approach is particularly valuable for Fanconi's anaemia because mismatched family transplants or transplants performed with HLA-identical unrelated donors have been disappointing, compared to better results with HLA identical cord blood transplants from family members.

This case demonstrates a high relevance of pre-pregnancy HLA matching together with preimplantation diagnosis for those conditions treated by identical HLA stem cell transplantation. So couples at risk of having a child with Fanconi's anaemia or other genetic condition, requiring HLA compatible stem cell donor, have now an option to initiate pregnancy not only free of disease, but also representing an HLA match for the affected sibling. Because only 1-3 embryos from 10-12 available in IVF on the average are usually selected for transfer anyway, pre-selection of these embryos based on their genotype seems to be perfectly acceptable on ethical grounds. It is understood that all the remaining unaffected embryos were frozen and are available for the couple, should parents wish to have more unaffected children, irrespective of their HLA type. In other words, the above work is not much different from the IVF procedure which routinely involves pre-selection of embryos based on morphological criteria, except the pre-selection process also includes the detection of mutation and the HLA type of the embryos, strictly relevant to medical indications.

The above case once again shows that preimplantation diagnosis has become an important part of assisted reproduction and genetic services, available for the growing number of patients in any community.