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Editorial
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Regression in the Implementation of Preimplantational Genetic Diagnosis in Spain Due to the Influence of Religious Views

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Editorial

Recently I examined a three month old girl that after genetic testing was confirmed to carry the same c.4269+1 G>T mutation gen NF1 associated with Neurofibromatosis as her mother [1,2]. The mother, presenting Neurofibromatosis phenotypically, had been diagnosed with this c.4269+1G>T mutation gen NF1 three years before her pregnancy. Unfortunately, this woman was not offered any type of Prenatal Genetic Counseling and therefore, she was not able to benefit from the Preimplantational Genetic Diagnosis (PGD). It is surprising to realize that this situation is very common for this and many other genetic diseases with potentially available preventive options.

The Spanish Law 14/2006 on Assisted Human Reproductive Techniques (ART) establishes under article 12 certain considerations for the application of PGD techniques. With the purpose of avoiding unnecessary formalities, this law allows performing PGD without the request of a case-by-case authorization for serious hereditable diseases with early onset and not susceptible to postnatal treatment (article 12.1.a) or to detect other alterations that could affect the pre-embryo viability (article 12.1.b).

This Law increases, however, the administrative control of the practice of ART and PGD with the nomination of the members of the National Committee for Human Assisted Reproduction (CNRHA). This law should avoid the freezing of policies, and at least, theoretically, facilitate CNHRA to adequately adapt to advances in science and current clinical practices. But the reality is very different. The uses of the great scientific advances achieved during the last decade in genetic testing, assisted human reproduction and stem cells are being intentionally restrained by the biased opinions of some members of the CNRHA, due to their personal religious views on these types of techniques and scientific research.

The advances in assisted reproduction and clinical genetics have allowed the development of preventive techniques with the use of PGD, a combination of *in vitro* fertilization, the biopsy of pre-embryonic cells, micromanipulation and techniques of cytogenetic and molecular diagnoses. The main purpose of PGD is to allow the analysis of pre-embryos in the lab after *in vitro* fertilization and before implantation in the maternal uterus. On day 3 post-fertilization, cleavage-stage pre-embryos of only 6-8 cells are biopsied to extract one cell for genetic diagnosis. Once the pre-embryo is confirmed to be free of specific hereditary abnormalities (such as mutations or chromosome alterations), it is then transferred to the mother's uterus. The use of PGD techniques do not lead to the alteration of any genetic content. The pre-embryo compensates for the absence of the biopsied cell and will continue to divide and develop normally.

Preimplantational genetic diagnosis could allow parents and doctors to prevent offspring from developing a long list of devastating monogenetic diseases, such as autosomal recessive (e.g. Cystic Fibrosis, Spinal Muscular Atrophy), autosomal dominant (e.g. Huntington's Disease, Type 1 Neurofibromatosis, Tuberous Sclerosis) and sex-linked (e.g. X-fragile Syndrome, Duchenne and Becker Muscular Dystrophies) (ESHRE PGD Consortium) Also, the presence of chromosomal reorganizations (Robertsonian translocations, reciprocal translocations and inversions) in one

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member of the couple could interfere with the possibility to conceive, increase abortions or congenic malformations. The use of PGD is also indicated in cases of alterations in the number and structure of the chromosomes.

In Spain, the first pregnancy after PGD happened in 1994 with the birth of a baby boy free of hemophilia from a couple where the mother was a carrier for the disease. Surprisingly, the CNRHA responsible for authorizing the cycles of PGD case-by-case when not corresponding to policies 12.1.a or 12.1.b, has not updated information about the practice of PGD in the country, neither the number of annual cases, nor the type of genetic conditions identified by the technique. The main reason for this lack of knowledge is the non-existence of a standardized protocol to collect data nationally. In the absence of such guidelines, the role of the CNRHA is obviously difficult when advising the medical administration.

The excessive formalities and waiting times that parents are submitted to before being evaluated case-by-case by the CNRHA is unacceptable. In addition, several very prevalent monogenetic diseases such as hereditary breast and ovarian cancer syndromes [3,4] associated to BRCA1 and BRCA2 mutations have to still be approved and accepted by the medical authorities in the CHRHA, some of whom are not supporters of the use of PGD techniques.

The first British baby genetically free from carrying the BRCA1 mutation by using PGD was born in 2009. However, up until today in Spain only four families have benefited from these scientific advances of assisted reproduction and PGD to prevent the passing of BRCA1 and BRC2 mutations. The first baby boy free of a BRCA1 mutation was born in 2011, and five years later, in 2016, another baby boy and two fraternal twins were born free of BRCA2 mutations. The first Spanish baby girl free of a BRCA1 mutation was born in August 2017.

Today with the high rate of success of PGD techniques, and the number of highly qualified Spanish biologists, embryologists and medical doctors substantiates that it is time to rethink whether the National and Ethical Committees are properly performing the function for which they were created [5]. Unfortunately, in Spain as well as in some other European countries, some religious organizations with more radical views have members in these Committees and are therefore involved in making very important medical decisions [6]. Furthermore, these members, with their individual partial/subjective points of view might be overly influencing and restraining the use in the 21st century of extraordinary scientific advances that could greatly benefit disease prevention and the quality of life of the current population and their descendants.

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