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# Selecting What? Pre-implantation Genetic Diagnosis and Screening Trajectories in Spain

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Genetic testing is one of the few biomedical sectors in which significant advances have been made in the past 20 years. Over this period of time, new genetic testing technologies have made their way into healthcare practices. Interestingly, the field in which most new genetic testing technologies have been introduced is human reproduction, especially assisted reproduction (Overall 2012; Rothman 2001).

Among these technologies, pre-implantation genetic diagnosis (PGD) and pre-implantation genetic screening (PGS) have raised enormous expectations due to both their ability to prevent the transmission of hereditary genetic diseases and their promise to improve success rates of in vitro fertilization (IVF) (Mastenbroek et al. 2007; Pehlivan et al. 2003). Often singled out as a paradigmatic example of soft eugenics, PGD has been criticized as yet another instance of the medicalization of reproduction, health and life (Ehrich and Williams 2010; Holm 2009;

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Krahn and Wong 2009). It has also been suggested that genetic testing, in general, and PGD, in particular, contribute to the commercialization and commodification of bodies, tissues and reproductive practices, in the context of a neoliberal strategy of individualization of medical risk and marketization of healthcare (Bunton and Petersen 2005). Experiences and imaginaries of patients undergoing PGD/PGS have also been explored (Ehrich et al. 2007; Lavery et al. 2002; Roberts and Franklin 2004), just as their political economy dynamics have been studied (Pavone and Arias 2012). However, most of these social studies have focused on PGD for molecular diseases while little work has been done on PGS.

This chapter aims at contributing to these debates by addressing what it means to go through pre-implantation genetic testing in Spain today. Spain alone performs almost half of all the PGD and PGS in Europe (Kupka et al. 2014). In Spain, however, PGS is, by far, the most implemented practice, representing more than 80 percent of the tests performed, according to a national registry that collects data from about 67 percent of the Spanish IVF centers (SEF 2013). Moreover, Spain has a very permissive legislation, which allows PGS to be performed without specific authorization, and adopts a set of very flexible criteria to grant permission to perform PGD. Finally, Spain has become a worldwide hub for assisted reproduction-related travel, both for PGD/PGS and egg donation, and possesses a remarkable IVF private sector, which features among the most technologically advanced in the Western world (Bergmann 2014; Salama 2014). Consequently, and contrary to the situation described in several other studies on PGD, the Spanish pre-implantation landscape is characterized by an unrestrained, massive use of PGS directed at increasing IVF success rates and a limited, stable recourse to PGD to positively select embryos free from specific genetic mutations or histologically compatible to their siblings for therapeutic reasons.

Drawing from interviews with patients undergoing PGD or PGS in different regions and clinics in Spain, we show that there is a significant difference in the experiences of women undergoing PGD and PGS. The experiences of women undergoing PGD with chromosome translocations—whose knowledge of their genetic condition followed their attempt to have children—lay somewhere between PGD and PGS, but are closer to the experiences of women undergoing PGS. These differences are not

(only) due to the different techniques, they are rather due to the different trajectories that the two techniques often entail. Many factors account for these differences, such as the clinical setting, referral routes and the actual health conditions. Though selection is at work in both cases, what is being selected and for what purpose is different for PGD and PGS, and so is the related experience. As such, examining how couples reflect on and experience embryo selection prior to implantation in Spain provides important insights into the material and conceptual intersections, convergences and blurring of assisted and selective reproduction in the twenty-first century.

## Selecting What? PGD, Reproductive Autonomy and Genetic Reductionism

Within reproductive medicine, pre-implantation genetic testing constitutes the technological core of the reprogenetics sector. It has been argued that it is a sociotechnical practice developed as part of a broader process re-configuring health, kinship and reproduction into increasingly medicalized practices within a general neoliberal strategy of market-based provision and regulation (Pownall 2013; Ruckert et al. 2015). Many studies addressing pre-implantation genetic testing wonder what it is that is actually being selected through genetic testing; what kind of biological characteristics are being given priority and what kind of social expectations, values and visions of life are being, thus, selected and reproduced. Indeed, pre-implantation genetic testing might be considered, in many ways, a family planning practice. As a consequence, it sparks questions about what kind of family types and relationships are being reproduced and what kind of society, and economy, is ultimately being endorsed. In order to answer these questions, different approaches to the study of genetics, health and society have been developed.

Some scholars have approached PGD and PGS from a perspective of biomedicalization of health, disease and identities (Clarke 2003; Clarke et al. 2010). From this perspective, PGD is seen as contributing to the ongoing transition from medicalization to biomedicalization, shifting the emphasis from enhanced control over external nature to the harnessing

and transformation of our internal nature (Ehrich and Williams 2010). Inspired by these studies, some have argued that pre-implantation genetic testing may be promoting a gradual shift from a complex, sociobiological view of human life to a narrow, reductionist approach where differences among individuals would be increasingly reduced to their genetic characteristics (Arribas-Ayllon et al. 2013; Bumiller 2009; Finkler 2011).

Other scholars have focused on the role played by genetic testing in a European context of increasingly privatized healthcare, constructed around patient choice models and inspired by new public management approaches. In this context, the increasing availability of genetic information is allegedly transforming the governance of population's health and encouraging an active participation of citizens constituted as "pre-patients", because of the genetic risk they carry, and as "potential consumers", for all the treatments they may have access to (Castiel et al. 2006). From this point of view, PGD emerges as yet another selective technique reinforcing the societal transition toward genetic welfare while consolidating an existing trend toward an individualization of health and care responsibilities.

Many studies have addressed the motivations of women and couples undergoing PGD. Some authors highlight the importance of reproductive choice, the desire to avoid abortion and the welfare of the future child as the main factors driving them to use PGD (Ormondroyd et al. 2012). Kalfoglou et al. (2005), for instance, argued that the use of PGD to avoid severe, life-threatening genetic illness or to select embryos that are a tissue match for a sick sibling was strongly supported, while its use to avoid adult-onset genetic disease, to select for sex, or to select for other non-medical characteristics was rather controversial. While these scholars have insisted that PGD for hereditary, life-threatening, genetic diseases increases reproductive choice and fosters individual autonomy, others have shown how this increase of choice opens a variety of more complex decisional scenarios (Järvholm et al. 2014). For instance, it has been observed that a history of miscarriages and infertility increases the willingness to undergo PGD, while the existence of an already affected child rather reduces couples' determination to use PGD (van Rij et al. 2011). It has also been argued that the increase of choice offered by PGD can actually hamper couples' choice (Zeiler 2004).

From a slightly different point of view, Hershberger and colleagues have focused on the importance of the individual psychosocial journey in the decision-making process (Hershberger et al. 2012). Similarly, Drazba et al. (2014) suggested that economic incentives and constraints play a crucial role in the decisional process. Others have rather stressed the importance of the IVF-stem cell interface (Franklin 2006), emphasising the higher propensity of women undergoing PGD or PGS to donate their "spare", genetically discarded embryos to research (Franklin et al. 2005; Svendsen and Koch 2008).

Finally, some scholars have studied the impact of national regulations, bringing to the fore how restrictive legislation may reduce the prescription of PGD in a particular national setting, while simultaneously encouraging a growing flow of affected couples going abroad to achieve it (Gianaroli et al. 2014), a phenomenon also known as reproductive tourism (Pennings 2002). It has also been argued that women undergoing PGD converge at the IVF clinic from a diverse range of reproductive and genetic trajectories, under very different psychosocial circumstances and with remarkably different worldviews (Karatas 2010). As already noted, the large majority of these studies have essentially focused on PGD for hereditary genetic disorders, while little is known about couples undergoing PGS.

## Aims and Methodology

This chapter aims at exploring and analyzing what it means to undergo both PGD *and* PGS in Spain today. Paying a special attention to the different *routinization* trajectories that PGD and PGS follow, the chapter collects and analyzes different experiences and perspectives of PGD and PGS patients.[1] What these two groups have in common is that embryo biopsies following IVF are used to select those embryos that will be implanted. This similarity notwithstanding, this chapter specifically addresses the following three main research questions: What are the motivations, expectations, doubts and concerns of women undergoing PGD and PGS? Are there relevant variations between PGD and PGS experiences? And what are the main factors that may account for these variations?

In order to address these questions, we conducted 21 semi-structured interviews with women undergoing PGD or PGS in private and public hospitals across different regions in Spain between 2010 and 2012. Originally gathered to investigate the articulation of PGD and PGS in Spain from the perspective of the women involved, these interviews were part of a broader data-gathering endeavor, which included interviews to policy-makers and medical professionals. Interviews were collected following the principle of maximum variation sampling (Creswell 2013) in order to ensure access to different experiences with regard to the technique, regional diversity and the type of healthcare setting (public, private or private with public subsidy). All women interviewed had undergone either PGD or PGS, or were considering doing it at the time of the interview. The interviews, organized around open-ended questions, explored different issues around PGD and PGS, such as personal experiences, choice of IVF center, level of information, access to the tests, psychological and genetic counseling, embryo donation, public versus private IVF settings and future prospects of the technique. The interviews lasted between 45 and 60 minutes, were recorded, transcribed and, finally, analyzed through a combination of *thematic data analysis* and *discourse analysis*. The former was employed to identify the most recurrent topics and to reconstruct the various organizational, medical and social steps of the PGD/PGS journey (Marshall and Rossman 2011). Discourse analysis, on the other hand, was used to identify, analyze and interpret the emotional, discursive and representational tools adopted by the interviewees to make sense of their experiences and to frame and communicate them to themselves and to others, including the interviewer (Potter 1997).

## Regulating Pre-implantation Genetic Testing in Spain

Although Spain's public healthcare system is well developed and highly valued by its citizens, assisted reproductive technologies (ARTs) and genetic testing are mostly accessed through the private sector (Pavone and Arias 2012). Indeed, the private sector has historically played a leading role in shaping the way in which ARTs developed in Spain, setting the agenda,

defining the problems that were likely to be addressed as well as the ways in which those were to be resolved (González 2014).[2] As a result, legal regulations followed, and were adjusted to, already existing practices, accommodating the needs and priorities of private clinics (Pavone and Arias 2012).

PGD was first regulated in 1988, when it was still an experimental technique (McClaren 1987). The 1988 Act considered pre-implantation genetic testing both as a tool to improve the success rate of assisted reproduction techniques and as a diagnostic tool for the detection of hereditary diseases. Given the experimental stage of PGD, the actual regulation was left to future legislative interventions based on three measures: the licensing and monitoring of authorized assisted reproduction centers; the setting-up of a consultative body to assess the government on the elaboration of appropriate legislative measures; and the creation of a National Registry, in which assisted reproduction activities could be recorded and stored (Alonso 2005).

In 1996, the law attributed the authority to license IVF centers to the regional governments but did not establish any specific authorization procedures for PGD and PGS. By then, private clinics had been performing these techniques without any specific regulation and control. The 1996 Bill, thus, entrusted the National Assisted Reproduction Committee (CNRHA) the regulation of PGD and PGS. Established in 1997, the CNHRA could not regulate the matter until 2006, when a specific bill introduced clear regulation criteria for PGD and PGS. The 2006 Act, however, did not establish a closed list of genetic conditions, introducing a more flexible regulatory regime to accommodate future technological advances without the need to modify the regulation. More specifically, the 2006 Act permitted the use of PGD for all the genetic hereditary conditions that could be considered "serious, early-onset and for which no treatment exist" and approved the use of PGD and PGS "to detect the alterations that may affect negatively the viability of the embryos". In all the cases that met these criteria, IVF centers and hospitals were expected to inform, through their regional authority, the CNRHA, from which no further authorization was required. All other cases required a specific authorization from the CNRHA (Muñoz 2012).

Nowadays, access to PGD is usually granted through public healthcare. Strict conditions apply to access the three cycles covered by the

social security system, and those are only available to women under 40. Recently, access to IVF treatments in the public healthcare system has been denied to single women and lesbian couples because "the absence of a male partner is not a medical condition"[3] (El País, July 18 and 23, 2013). Yet, some regions contested this measure, refusing to implement it. The public healthcare system, however, implies long waiting lists of more than two years, depending on the regions (Adeces 2015). While in a traditional IVF, couples with living offspring are prevented from accessing subsidized assisted reproduction; this condition does not apply to couples undergoing PGD to avoid transmission of hereditary diseases. Finally, being a controversial technique, whose effective ability to improve success rates has been extensively criticized (Hardarson et al. 2008), PGS has not been included in the social security system. While it has never been at the center of any public debate or controversy in Spain, PGS remains accessible only in private clinics where it is generally offered as an extra service enhancing the chances of success of the IVF cycle, which very much reveals the intense commercialized approach in the Spanish political economy of ARTs (Pavone and Arias 2012).[4]

## Main Findings: Pre-implantation Trajectories

In 2014, Sociedad Española de Fertilidad (Spanish Fertility Society) (SEF) reported 2890 PGDs and PGSs. Of these, about 1000 were PGD performed on molecular diseases, cytogenetic diseases or specific chromosome translocations associated to miscarriages, while the rest were PGS performed in relation to advanced maternal age (almost half of all the tests performed), and repeated implantation failures (see Table 6.1).

**Table 6.1** PGD and PGS distribution—Spain 2013

| | |
|---|---|
| Molecular diseases (PGD) | 340 |
| Cytogenetic diseases (PGD) | 373 |
| Miscarriages (PGD) | 340 |
| Advanced maternal age (PGS) | 1194 |
| Implantation failure (PGS) | 222 |
| Others | 305 |
| **Total** | **2890** |

*Source*: SEF 2014

A key finding of our study is that the experiences and trajectories of the women interviewed vary significantly depending on whether they have undergone PGD or PGS. This variation is partially related to the fact that the two techniques are very different and pursue different purposes. Both PGS and PGD are used in efforts to overcome biological obstacles to reproduction (assisted reproduction), while only PGD is used in efforts to avoid transmission of a known hereditary disease or to create a so-called savior sibling (selective reproduction) (cf. Gammeltoft and Wahlberg 2014). Whilst some PGDs are performed to look for specific chromosomal translocations, most of them look for specific genetic mutations in alleles, mostly through PCR (Polymerase Chain Reaction). PGS, in contrast, usually consists of a karyotype, mostly performed through a FISH study (fluorescence in situ hybridization), looking for both chromosome numerical and structural abnormalities. The variation is also due to the fact that PGS is only offered and accessed in the private sector, while PGD can be accessed in both healthcare settings. Finally, part of the variation is due to the different trajectories women using PGD and PGS follow, as they converge at the IVF clinic from different journeys, under different circumstances and for different purposes.

Importantly, we found a high degree of confusion between the two techniques in patient narratives. The two techniques, known as *diagnostico genético pre-implantacional* (PGD) and *cribado genético pre-implantatorio* (PGS) in the clinics are both translated as DGP, *diagnóstico genético pre-implantacional*, that is, both are identified by the same name, blurring the differences between the two. For instance, women often wondered why for some people it was more *difficult* to get access to a technique that seems rather easy to access for others. They also wondered why it was covered by the public system in some cases but not in others, suggesting that they were not aware of the existence of two different techniques, both named "DGP".

> I never actually got to understand, because in all the websites, in the media, everywhere it is written that PGD can only be accessed if certain requirements are fulfilled and after the authorization of the Ministry… but we were having PGD without any authorization or any special requirements… you see what I mean? […] I never understood this. I don't understand why

> some couples need authorization, other couples who did not get authorization went to other countries… the fact is that I was having PGD without going to the public hospital, and without having to ask for permission…I was confused, but given that the only thing I wanted was to have a child, I ultimately did not care. [P16]

In spite of this blurred distinction between the techniques, the interviews reveal the existence of three different experiential trajectories: one for PGS and two for PGD. Of these latter two, women who had PGD performed for chromosomal translocation show an experience quite similar to the women undergoing PGS, as both groups were pursuing assisted reproduction. These women were, in fact, offered PGD without having previous knowledge of their genetic conditions: their journeys to IVF, and consequently to PGD, were linked to recurrent miscarriages, eventually diagnosed as linked to chromosome translocations. The experiences of women undergoing PGD as selective reproduction to avoid transmission of molecular, hereditary diseases, in contrast, are very different from the previous two. Consequently, our full account of these differences in the following will show how PGS women have experiences very similar to those identified in the literature about ART patients' trajectories (Cussins 1996; Friese et al. 2006), while women undergoing PGD for genetic disorders have experiences more similar to the ones discussed in the literature on selective reproductive technologies (SRTs) and on PGD elsewhere in Europe (Franklin and Roberts 2006). Interestingly, even if women undergoing PGD for chromosomal translocations are pursuing assisted reproduction, their experiences contain key elements from both of the other two trajectories (Table 6.2).

## Women Undergoing PGS

The journey of the women undergoing PGS begins with a fertility problem: they cannot get pregnant or the pregnancy does not get to term (either the embryo does not implant or ends up with a miscarriage):

**Table 6.2** Three different pre-implantation trajectories

| Issues | PGS | PGD ct | PGD md |
|---|---|---|---|
| Main trigger to IVF | Infertility | Miscarriages or implantation failures | Hereditary genetic condition |
| Frame of the technique | Tool to improve IVF success rate | Only way to get pregnant | Prevent birth of affected offspring |
| Clinical setting | Private clinics | Both private and public hospitals | Both private and public hospitals |
| Level of prior information | Low | Medium-low | High |
| How they get to know about the technique | Offered by the clinic | Previous diagnosis | Previous diagnosis or affected offspring |
| Alternative to abortion | No | Occasionally | Yes |
| Main purpose | Getting pregnant | Have a successful pregnancy | Have a child free from a specific disease |

> Whenever I got pregnant I had some problems and it never worked out in the end… there was a genetic problem… not one affecting the egg, rather one proceeding from the assemblage of the egg and the sperm. I wanted to go for a safer and more effective option because my womb was being damaged… and you know I need my womb to become a mother [P12].

These patients are usually offered PGS as a tool to increase the chances of success of IVF, using embryo biopsy to choose among embryos. Framed as an effective technique to enhance success, some women suggested that the public sector should include it to improve cost-effectiveness: "*The public healthcare system would be better off because maybe with one cycle only* [IVF with PGS]" or "*you can have more success than with three normal cycles* [without PGS]" [P12]. Nonetheless, the relatively low success rate often generates second thoughts, disappointments and frustration.

> We thought it was something almost foolproof, but then, once in the clinic, we realized that, with so many people there, it wasn't so foolproof, it is just one more option, that gives you much more opportunities with high

> technology, but clearly it is not foolproof and clearly that came to me as a disappointment [P6].

Both patients and professionals are aware that PGS is a tool for selecting better embryos but "better" is understood in several, ambiguous ways. It is said to refer to a *correct* chromosome profile of the embryo, but it also implies the idea of "good quality" and "healthiness":

> PGD [in this case PGS] analyses each and every embryo in order to detect the ones that are perfect from a chromosome point of view and can survive better. Sure, it is also true that PGD can negatively affect the embryos but, then, those embryos that survive PGD are the ones that are most likely to continue to term. [P9]
>
> I know that [PGS] analyses some chromosomes and then helps selecting those that are healthy, genetically healthy […] given it was our last chance, we thought that selecting a healthy embryo would give us more chances. [P8]

These women associated higher chances of success with selection of "health(ier)" embryos, referring to them in abstract terms or understanding "healthy" as "endowed with ability to develop to term". It is precisely this encouraged conflation between "chromosome normality" and "ability to develop to term" what moves patients into undergoing PGS, expecting it to raise their chances of success in the assisted reproduction journey.

> As it was our last opportunity, to select a healthy embryo to have more chances… to be honest, well, the doctor told us that with two healthy embryos there is 50 per cent of likelihood, which is not much if you consider the cost, but well… [P8]

In one case, PGS is mentioned as a tool to get rid of "bad" embryos due to potential genetic conditions. Here, the genetic quality of the embryo was used by the clinic to persuade the patient to try IVF with PGS.

> I had to go for IVF with PGD in order to get rid of the embryos that were "bad", so to speak […] they told me that, well, that it had to be done with pre-implantation diagnosis to, well, to… to put the healthy embryo

> because there were not healthy ones, so they explained a little bit of the technique to me, well, they explained everything to me. [P12]

In the PGS trajectory, selection procedures are not framed in terms of specific alterations or mutations but in terms of a quality assessment procedure identifying and separating—on the basis of chromosome characteristics—"good" embryos from "bad" embryos. Health is geneticized: embryos are considered healthy or unhealthy on the basis of their genetic profile, interpreted by pre-implantation screening.

> In our first clinic, they told us 'well, you have high chances of having children with anomalies' like Down syndrome, Turner syndrome … Then obviously with this technique in principle before being implanted they analyzed the embryos before and they would only place on you the "good ones", I mean, the ones that were genetically healthy. [P16]

The fact that these women end up in PGS is entangled in a longer history of being immersed in ARTs, blurring once again the differences between ARTs and SRTs as long as their trajectories come to matter. The experiences these women told us were highly framed by their need for *assistance* on their reproductive endeavor and selection *per se* only came to be relevant as long as genetic factors were used as an explanation to their reproductive problems. Thus, these women were having their embryos selected as a way of increasing their chance of having an actual baby, and even if selection was linked to genetics and took place at the embryo level, their experiences seem not to be that different to those of other women undergoing ARTs without pre-implantation genetic testing, whose embryos are selected on the basis of the visible, morphological features of the developing embryos rather than on their chromosomal configuration.

## Women Undergoing PGD with Chromosome Translocation

The trajectory of women undergoing PGD for chromosome translocations stands somewhere in between those related to genetic PGD and those related to PGS. In this case, PGD targets specific chromosome

translocations and anomalies, focusing on previously identified hereditary chromosome alterations. Women undergoing have a degree of information, awareness and knowledge that is generally higher than the one observed in the PGS group and they do not frame PGD as a tool to boost their chances to get pregnant, but rather as "the only alternative" to achieve their desired motherhood.

> The repetition of miscarriages was somehow due to the inversion of the chromosome, and so for us to achieve a pregnancy we had to do an in vitro fertilization and, after that, we have to do an embryo selection […]. There was not much alternative. [P10]

Women in this group both use and resist the narrative of selection. They do so by re-framing the idea of selection, insisting that they are not selecting embryos on the basis of personal criteria and emphasizing that they are giving a chance for healthy children to be born. Thus, selection is considered legitimate under a discourse of health and pregnancy success but rejected on the basis of any other criteria (e.g., choosing gender or any other characteristics of the phenotype).

> You don't look to see if it is a boy or girl, you are not looking for sex selection or discarding healthy embryos…it is more… more like the opposite, as long as they are healthy…[everything is good]. [P7]

Women undergoing PGD for chromosome alterations converge at IVF clinics mostly because their pregnancies cannot develop to term. In their case, the genetic component is both relevant and specific, and encourages them to further "geneticize" their reproduction experience, reducing their fertility problems to chromosome translocations.

## Women Undergoing PGD for Molecular Diseases

The third group of women accessed PGD as a result of hereditary genetic conditions. They do not have fertility problems, but many have lived with a close relative affected by a genetic disease and opted for PGD as

a particular way to become parents. PGD is their fundamental reason to choose IVF because they want to prevent their progeny from developing the same disease affecting them or their close relatives. Some have already had children who have inherited the disease. In fact, two out of three kids born to these women before the interview were deceased at the moment of the interview, while the third one was under chemotherapy treatment. Clearly, in such cases, PGD is framed as a selective device enabling parents to have offspring free from hereditary diseases.

> Both my husband and I, we are carriers of a genetic disease. We had a baby who developed the disease and died when he was five months old. Then, we decided that we wanted to have our second baby through PGD. [P23]

In these cases, women actively seek PGD, and often they get to the clinics with a deep and extended knowledge about their disease, the techniques, risk of transmission and the chances of success.

> When I went to the gynecologist, I already knew that they offered this technique, I knew that I had to go through it … in other words, it wasn't the doctor, it was me who already knew because I had seen it in the internet and in the press, when the first cases of children free from the disease were announced. [P19]
>
> We had a fifty per cent chance that the baby would be born with the disease, so we decided to go the clinic. [P25]

In their narratives, healthy is no generic term: it means free from a specific hereditary disease.

> I… well I have a genetic problem, a genetic disease that I share with nearly all the women in my family, it is inherited from my father […] Given that I wish to have children, I would like to have them healthy…free from this disease, I mean. [P27]

PGD is more often framed as a medical tool helping children to come to life free from a disease, rather than as a tool to help parents to have kids free from it, even if both forms of reasoning are present. In other words, PGD is presented more in relation to the child than to the parents.

> My mother has got a neurodegenerative disease, and this is hereditary so, a few years ago, the whole family was tested and it turned out I was a carrier […] if I have children there is 50 per cent chance that they develop the disease, which is a fatal disease for children […] Therefore, for me this is clear, that if there is anything I can do to ensure that my children will be healthy, I will do it. [P26]

As a result of this unique connection with the health of the prospected children, PGD is endowed with a special moral and social status, making it the only acceptable way of fulfilling their reproductive project:

> My father and my son have a hypertrophic myocardial pathology and when I went for a genetic test and it came out positive, the doctors suggested I should go for pre-implantation genetic diagnosis […] when this was suggested to me, I no longer considered having a child in a different way. [P13]

These women generally considered PGD an alternative to abortion. Indeed, some turn to PGD after difficult experiences trying to have children through regular pregnancies in combination with prenatal testing, as recommended by their doctors.

> The geneticist in the Hospital [omitted] had no idea, he just suggested I should get pregnant in a normal way and, then, if the retinoblastoma would be detected in the fetus, I could always interrupt the pregnancy, which was legal to do. He never mentioned the possibility of selecting the embryos, which would have been way less painful. [P22]

While their medicalization dynamics are different, these women adopt patient logics more than the women in the two other groups. They present themselves as willing to undergo a medicalized reproductive process so as to prevent their children from going through the kind of medical journey their relatives have experienced. Thus, they accept becoming patients to avoid their children doing so. In their narratives, the experience of being close to patients is the key factor determining their reproductive trajectory:

> If we had to have a child again, we would run the risk of going through this again, because both my husband and I, we have a mutated gene so…[…], after I had my son sick from this disease, I know this disease, it is atrocious […] I do not want to have another child going through this again… this is a terrible disease. [P20]

The technique is not framed in individual terms nor is it linked to the desire of having children, as they are not going through PGD to become parents. Instead, PGD is framed in generational terms, for they are going through it to ensure that the next generation will be free from a disease that has haunted their family.

> The fundamental reason to opt for PGD was that I have seen my father and my brother, and I know the quality of life they have, and I guess that all the people who share this kind of disease know how it dramatically affects the quality of life of those who are closer to you, and then… you don't even think this could be a possibility. [P13]

The logic of selection adopted is articulated around two main elements: on the one hand, preventing their children from suffering (or dying) and, on the other hand, preventing themselves from having to go (again) through the suffering they experienced with their affected relatives.

> I believe that [PGD] offers an opportunity to get rid of diseases that have been devastating the quality of life of many people till now… it would be great if PGD would also give this opportunity in relation to more diseases… but at least it allows us to avoid some diseases…It would be nonsense not to take advantage of this opportunity. [P13]

Ultimately, the trajectories of these women are significantly built around the issue of selection, confirming thus many of the findings discussed elsewhere in the literature on PGD. Here, the fact that their reproductive processes are assisted technically is a means to one particular end: enabling the selection of embryos free from specific and known diseases. Interestingly, this specifically medicalized reproduction route is framed as a temporary way of escaping medicalization in the long run (for their potential children).

## Conclusion

This chapter has focused on how PGD and PGS are experienced in Spain today. While most of the studies on PGD tend to focus on the individual experiences of women undergoing PGD for hereditary molecular diseases, we have included in this study women undergoing PGD for cytogenetic diseases and chromosomal abnormalities as well as women undergoing PGS. We interviewed 21 women undergoing PGD and PGS in public and private hospitals across different regions in Spain, with the aim of casting light on the differences and commonalities in the experiences and trajectories of the women undergoing these techniques.

Our analysis shows that there exist relevant differences between the experiences associated with these techniques and the approaches to selection by the related patients. Women undergoing PGS and PGD for chromosome translocations consider the technique a tool to boost their chances to achieve a successful pregnancy, seen as their only chance to become mothers. In that sense, the techniques seem to engender experiences similar to those recorded and discussed in previous studies of women undergoing IVF (Cussins 1996; Friese et al. 2006). The medicalization process, however, differs: PGS targets broader infertility while PGD addresses specific types of chromosomal translocations only. The selection logic in both cases is applied to discard "bad" embryos, understood as those which would not develop further, and select "healthy" embryos that are expected to be able to survive until birth. In contrast to those two trajectories, women undergoing PGD for molecular diseases accept to undergo medicalized reproductive processes to prevent their offspring from developing a disease that has been haunting their families. These women have vivid experiences of the disease, either because their first child developed it or because close relatives are affected. As a result, the selection logic is different: they do want to select "healthy" embryos, but healthy does not mean "fittest" but rather "free from a specific disease". Following the distinction between assisted reproduction and selective reproduction made in this volume, we can now clearly speak of an SRT trajectory, in which selective reproduction is the key force driving these women to undergo the whole procedure.

Importantly, the information these women possess and receive is different. Often those undergoing PGS do not have information about the kind of selection their embryos are subjected to. They know that their embryos go through a process of genetic selection but they might not know how this process works exactly. Indeed, some are not aware that there are two different techniques, both abbreviated as DGP in Spanish. They are not that interested in knowing which kind of chromosomal issues are being screened out, as long as the selected embryo has more chances to result into a baby. They distinguish between healthy and unhealthy embryos assuming unhealthy to be "not likely to live to birth". In PGS, selection is, thus, a means to an end, another assisted reproduction technique meant to help traditional IVF to succeed. The trajectory of women undergoing PGD for translocations or miscarriages is, in many ways, similar to this PGS trajectory. Some of them are more aware of the particular genetic problems they may have and may also be more informed about the techniques, but they look for PGD for the same purpose: a successful pregnancy. In contrast, women who access PGD in order to have offspring free of a specific genetic condition follow a different trajectory. They arrive informed; they have been through either selective abortion(s) or they live, or have lived, with family members affected by a disease, and they know pretty well what kind of selection procedure they need and for what specific mutation. Thus, PGD is the actual end, and IVF is the means to it. Their narratives differ from those of women with fertility problems, even if their genetic conditions bring them to the same clinic (which is not always the case, as PGD is offered by private as well as public hospitals).

These results suggest that selection is always at work but that the experience of selection is different. They also suggest that there is no clear boundary between ARTs and SRTs, and that PGD/PGS contribute to the further blurring of a boundary that has always been blurred. IVF, and ARTs in general, might have always been about selection and PGS works here as yet another example of it. In IVF, selection of gametes and embryos is done by using the technology of the informed, trained and selective gaze of the embryologist who either chooses gametes or ranks embryos in A, B or C quality according to their (observable) ability to multiply and survive. PGS is lived by these women, in a way, as a

technique extending the scope and power of this selective gaze into the chromosomal domain of the embryos but maintaining the same purpose. PGD extends it further down into the gene's structure and, allegedly, maintains and extends this purpose, too, by screening out specific genetic mutations to prevent the birth of individuals carrying (the risk of) specific genetic diseases. The experience of the women, nevertheless, showed that *selection*, in this second sense, is the real driving force in very few specific cases, which represent the minority of the PGD that takes place in the Spanish context.

This chapter makes a contribution to the existing literature by showing that these two techniques in Spain are embedded in different medical contexts and trajectories. The differences in the experiences we have collected are not (only) due to the technique per se, but rather to the different trajectories within which they develop. Many factors account for these differences such as the clinical setting, the actual health conditions of couples and also their previous reproductive trajectories. What we would like to emphasize is that two different types of selections are at stake in PGD and PGS: what is being selected and for which purposes is different, and the trajectories of the women may have more of an impact on what selection means than the technique itself, as the experiences of the second group of women show. While our results are consistent with many other studies on PGD for genetic mutations and diseases, we do need to emphasize that PGD for molecular disease is not what effectively characterizes the Spanish PGD landscape. The latter is rather characterized by an unrestrained, massive use of PGS as an assisted reproduction technique whose primary aim is an increase of the success rate, and by a limited, stable recourse to PGD to positively select embryos free from specific genetic mutations or histologically compatible to their siblings for therapeutic reasons.

## Notes

1. Routinization has been defined as a "socio-historical process whereby certain forms of medical technology come to be (re)produced and entrenched within particular juridical, medical, social, economic, cultural and institutional configurations" (Wahlberg 2016: 98).

2. While in the late 1980s, there were only 14 IVF centers, today 200 IVF centers exist in Spain, 165 of which are private and 35 of which are public SEF. 2013. *Registro de la Sociedad Española de Fertilidad: Técnicas de reproducción asistida (IA y FIV/ICSI)*, 1–41. Madrid: Sociedad Española de Fertilidad.
3. El País Website, http://sociedad.elpais.com/sociedad/2013/07/18/actualidad/1374178125_262676.html, last accessed 20 May 2015, and http://sociedad.elpais.com/sociedad/2013/07/23/actualidad/1374575386_841886.html
4. In our previous round of interviews (2008), some embryologists and gynecologists admitted that new evidences was clearly showing a reduction of success rates associated with PGS and forecasted a decrease in the use of this technique. However, the most recent data (2013) show no sign of decline but rather a marked increase. In 2009, 1037 PGS were performed over a total of 1683 PGD/PGS and 40,704 IVF cycles. In 2013, 2064 PGS were performed over a total of 2890 PGD/PGS and 46,911 IVF cycles. While in 2009, PGS for advanced maternal age accounted for 26 percent of all PGD/PGS, this percentage rose to 41 percent in 2013.

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