To Avoid Passing A Faulty Gene, Genetic Diagnosis Is Key

By Ana-Marija Dolaskie — March 9, 2016

When singer John Legend and wife Chrissy Teigen used PGD— or pre-implantation genetic diagnosis [2], to choose the sex of their baby, social media went nuts [3]. The prenatal gender decision is one of the optional add-ons that can be performed through PGD, and it is offered by fertility clinics to hopeful parents who undergo IVF treatments. The procedure is controversial, and the couple took some heat; after all, Tiegen did not conceive a baby girl by chance.

But when the procedure is used for its true intent, to detect and prevent the transmission of faulty genes carried by one—or both—parents, is the controversy still there? Maybe, maybe not.

The technique has been around since the early 90s, first being used only in cases for cystic fibrosis. Now, it is approved for more than 250 conditions [4]. But the requirements for couples are strict: If there is a confirmed genetic condition from either parent or family, with a significant risk of transmission to baby (more than ten percent), couples can get funding for the procedure. Once approved, couples undergo IVF treatments to create embryos, which are then analyzed for rogue genes, a process called pre-implantation genetic screening (PGS). The embryos that are clear of those genes are then implanted in the womb. Couples who already have a child without the specified condition would not qualify for the procedure.

And in the case of the BRCA1 or BRCA2 genes, the decision, for some, isn't clear cut. Although both genes are approved by the HFEA [4] (Human Fertilization & Embryology Authority), some doctors aren't sure the quite involved PGD procedure is justified.

'Pre-implantation genetic diagnosis was initially developed for very specific life-threatening conditions that a child would definitely get. But using it to avoid BRCA 1 and 2 mutations effectively screens out embryos who might never suffer from that disease. When you're offering it for the risk of disease rather than the disease itself, that becomes a more open-ended question," Dr Anna Smajdor, a lecturer in medical ethics at Norwich Medical School, told the Daily Mail [5].

According to the latest estimates [6], 55 to 65 percent of women who inherit the BRCA1 mutation and around 45 percent of women who inherit the BRCA2 mutation will develop cancer by the age
of 70. For men who carry either gene, the risk of prostate cancer is increased. So for Paula Macrae, who carries the BRCA2 gene (and her mother has battled both breast and ovarian cancer), the choice was clear. The alternative would have been not having children at all, she told the Daily Mail. [5]

PGD has the potential to not only select embryos without a genetic disorder, but it can also increase chances of pregnancy, it can match a sibling in HLA type [7] in order to be a donor, it can help reduce cancer predisposition, and—in the case of John Legend's soon to be born baby girl, it can also help with gender selection. Undoubtedly, the ethical concerns are there; but if the advancement in medicine has come this far, are we crazy not to embrace it?

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