WANTING BABIES LIKE THEMSELVES, SOME PARENTS CHOOSE GENETIC DEFECTS

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Wanting to have children who follow in one's footsteps is an understandable desire. But a coming article in the journal Fertility and Sterility offers a fascinating glimpse into how far some parents may go to ensure that their children stay in their world — by intentionally choosing malfunctioning genes that produce disabilities like deafness or dwarfism.
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The article reviews the use of preimplantation genetic diagnosis, or P.G.D., a process in which embryos are created in a test tube and their DNA is analyzed before being transferred to a woman's uterus. In this manner, embryos destined to have, for example, cystic fibrosis or Huntington's disease can be excluded, and only healthy embryos implanted.

Yet Susannah A. Baruch and colleagues at the Genetics and Public Policy Center at Johns Hopkins University recently surveyed 190 American P.G.D. clinics, and found that 3 percent reported having intentionally used P.G.D. "to select an embryo for the presence of a disability."

In other words, some parents had the painful and expensive fertility procedure for the express purpose of having children with a defective gene. It turns out that some mothers and fathers don't view certain genetic conditions as disabilities but as a way to enter into a rich, shared culture.

It's tempting to see this practice as an alarming trend; for example, the online magazine Slate called it "the deliberate crippling of children."

But a desire for children with genetic defects isn't new. In 2002, for example, The Washington Post Magazine profiled Candace A. McCullough and Sharon M. Duchesneau, a lesbian and deaf couple from Maryland who both attended Gallaudet University and set out to have a deaf child by intentionally soliciting a deaf sperm donor.

"A hearing baby would be a blessing," Ms. Duchesneau was quoted as saying. "A deaf baby would be a special blessing."

Born five years ago on Thanksgiving Day, the couple's son, Gauvin, was mostly deaf, and his parents chose to withhold any hearing aids.

Controlling a child's genetic makeup, even to preserve what some would consider a disease, is the latest tactic of parents in an increasingly globalized society where identity seems besieged and in need of aggressive preservation. Traditionally, cultures were perpetuated through assortative mating, with intermarriage among the like-minded and the like-appearing.

Modern technology has been adopted for this purpose; for example, a quick Web search reveals specialized dating services for almost any religious or ethnic subgroup. Viewed in this context, the use of P.G.D. to select for deafness may be merely another ritual to ensure that one's children carry on a cultural bloodline.

Still, most providers of P.G.D. find such requests unacceptable. Dr. Robert J. Stillman of the Shady Grove Fertility Center in Rockville, Md., has denied requests to use the process for selecting deafness and dwarfism. "In general, one of the prime dictates of parenting is to make a better world for our children," he said in an interview. "Dwarfism and deafness are not the norm."

Dr. Yury Verlinsky of the Reproductive Genetics Institute in Chicago, who also refuses these requests, said, "If we make a diagnostic tool, the purpose is to avoid disease."

But both doctors said they would not oppose sending families to other doctors who might consent.

Today, parents increasingly use medical procedures to alter healthy bodies. In 2003, for example, the Food and Drug Administration

granted approval to Eli Lilly to market human growth hormone for "idiopathic short stature," or below-average height in children — to make them taller, purely for social reasons. Theoretically, almost a half million American boys qualify for treatment. Why, some may argue, should choosing short stature be different?

Mary Ellen Little, a New Jersey nurse with dwarfism, had her first daughter before a prenatal test for achondroplasia was available. For her second child, she had amniocentesis. "I prayed for a little one," meaning a dwarf, she told me.

The wait, she recalled, was grueling, since "I figured I couldn't be blessed twice, but I was." Both her daughters, now 11 and 7, are "little people."

The major barrier to Ms. Little's simply choosing her children's height is ease. To her, P.G.D. to select for dwarfism is too invasive; however, if having dwarf children were simply a matter of trying to conceive at a certain time of the month or taking a pill, she said, "I would do that."

Barbara Spiegel, a homemaker in Maine who has dwarfism, had a first pregnancy that ended in miscarriage. She underwent genetic testing during her second pregnancy, and because of a laboratory mix-up involving petri dishes, was told that her child would grow to normal height. She would have loved the child, she said, but in an interview, she recalled thinking, "What is life going to be like for her, when her parents are different than she is?"

She worried that the child would be teased excessively. Ms. Spiegel's best friend, who has average height, has a daughter with dwarfism, and the child sometimes comes to Ms. Spiegel for support; maybe an average-size child would also go to others for motherly advice. For a brief time, Ms. Spiegel grieved because she felt a dwarf baby would have been "just precious." But after a week, the mix-up was detected and she got her wish.

Genetic testing for dwarfism has an extra ethical wrinkle. When both parents are dwarves, their embryos have a 25 percent chance of normal height, a 50 percent chance of dwarfism, and a 25 percent chance of what is called a double dominant mutation, which is usually fatal soon after birth. Because many dwarf mothers worry

that their fetuses might have the fatal mutation, those who conceive without assistive technology, like Ms. Little and Ms. Spiegel, often undergo amniocentesis or chorionic villus sampling to detect double dominant mutations. Many consider abortion if the test is positive — but many would carry either a dwarf or an average-height child to term.

Preimplantation genetic diagnosis can identify embryos with double dominant mutations, so they can be discarded before implantation, while preserving embryos destined for either dwarfism or average height. In dwarves, then, P.G.D. could help avoid many doomed pregnancies if double dominants were never implanted. But then a choice would have to be made, since the genes are known. And many dwarves might select embryos for dwarves — although others might choose those for average-size children.

Dr. Stéphane Viville, who first reported P.G.D. for dwarfism in 2003 in France, used it to eliminate embryos with dwarfism among couples where one member was a dwarf and the partner had average height. Interestingly, if confronted with a situation where both parents were dwarves, Dr. Viville says that he most likely would implant only an embryo destined for normal height — and forbid not only double dominants but also dwarf embryos.

I think Dr. Viville fears that P.G.D. could be used willy-nilly to make genetic freaks. Yet the same fears pervaded the issue of in vitro fertilization decades ago. The small number of P.G.D. centers selecting for mutations doesn't bother me greatly. After all, even natural reproduction is an error-prone process, since almost 1 percent of all pregnancies are complicated by birth defects — often by more disabling conditions than dwarfism or deafness.

More important, as a physician who helps women dealing with complex fetal diseases, I've learned to respect a family's judgment. Many parents share a touching faith that having children similar to them will strengthen family and social bonds.

Of course, part of me wonders whether speaking the same language or being the same height guarantees closer families. But it's not for me to say. In the end, our energy is better spent advocating for a society where those factors won't matter.

Related Question: What would you do if you were a doctor being asked to perform PGD to choose a child to be born with genetic defects?