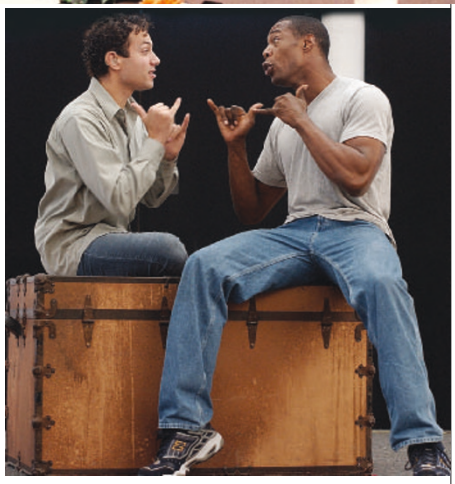
DEAF by design

Employing genetic diagnosis to avoid having a baby with a disability is controversial enough. But a minority of deaf people would consider testing to ensure that they had a deaf child. Carina Dennis finds out why.

S. PERLMAN/AP



G. DOMENICO/AP

All together now: deaf culture now encompasses everything from spelling bees (audience shown applauding, above) to Broadway shows (right).

John and Karen — not their real names — are both deaf, and desperately wanted a deaf baby. But genetic testing showed that this was extremely unlikely. "They were devastated," recalls Arti Pandya, a clinical geneticist at Virginia Commonwealth University in Richmond, who counselled the couple. It was two years before they got over their disappointment and started trying to conceive their first child.

The couple's attitude will shock many people. If you can hear, it's hard to understand why anyone would want a deaf child. But John and Karen's views are not that unusual among those who identify themselves as 'Deaf' with a capital 'D'. The Deaf view their condition not as a disability, but rather as the underpinning of a rich culture that should be celebrated and preserved. And with the identification of the most common genetic mutations linked to deafness, it is now possible, in theory, to make an active choice to have a deaf child.

This possibility turns the debate over designer babies on its head, providing ethicists and genetic counsellors with a dilemma. Only a tiny minority of deaf people would wish to use genetic tests in this way. Some argue that their reproductive choices should be respected. But is society prepared to sanction the use of genetic diagnosis for a purpose that many find difficult to understand — and some might even see as immoral?

Some Deaf people despair of ever being understood by those who aren't part of their culture. The Deaf identity is in large part a product of a shared sense of isolation from the hearing world. "Exclusion is central to the experience," says Gary Kerridge, regional disability liaison officer at the University of Ballarat in Mount Helen, Australia, who lost his hearing as a young child.

For deaf children, the majority of whom are born to hearing parents, even family gatherings can be lonely affairs. Many of them feel liberated by their first experience of Deaf culture. "They learn to sign and suddenly for the first time, after years of being isolated and struggling, they are accepted," says Kerridge. "Naturally, they quickly develop a strong attachment to the Deaf way of life."

A world of their own

Sign language is central to the lifestyle. It uses hand shape, position and movement, plus posture, facial expressions and other visual cues, to form words and convey meaning. It has its own rules for grammar, punctuation and sentence order. It is elaborate and expressive, and lends itself readily to poetry and theatre.

For a hearing person, entering a room full of chattering signers can be disconcerting. Methods used to attract attention, for example, seem downright rude. "Stomping on floors, waving animatedly, flashing lights and thumping tables are all considered OK," says Kerridge.

Knowing sign language doesn't, by itself, break down the barriers between the hearing and the Deaf. "Even hearing people from Deaf families and who sign well are always, to a certain degree, seen as culturally distinct," says Kerridge. "That absolute feeling of exclusion from the hearing world is difficult for a hearing person to fathom."

Within Deaf culture, however, there's a level of social intimacy that is rare among the hearing. "I will meet another Deaf person for the first time and in five or ten minutes, it's not uncommon to know a great deal about their family and personal life," says Carol Padden, a linguist at the University of California, San Diego, who was born deaf, to deaf parents. "I have to remind myself not to expect the same invitation to become familiar when I'm with hearing colleagues."

That, in a nutshell, is why some deaf couples would prefer to have deaf children. Communication and the pursuit of intimacy are central to being human. If you genuinely believe that your children will have at least as rich an emotional life if they cannot hear, and

M. KOOREN/REUTERS

Good vibrations: the deaf community's experiences, such as 'listening' to a concert through a balloon, can be difficult, if not impossible, to explain to the hearing.

that you will be better able to communicate with them, why not make this choice?

"I don't see anything wrong with it. I see it as being similar to how parents determine the religion or education of their child," says Ted Supalla, who has been deaf since birth, and studies sign languages at the University of Rochester in upstate New York. Supalla's own children can hear; they communicate with him by sign language and speak to his hearing wife.

Genetic lottery

Like Supalla, most deaf people are happy to let nature take its course, and say that they would be content to have a hearing child. But deaf people are increasingly marrying one another, making deaf children more likely. A report published in April theorized that the increasing number of marriages among the deaf during the nineteenth century may have doubled the frequency of deafness in the United States caused by mutations in genes for proteins called connexin 26 and connexin 30, which affect the function of the ear's sound-sensitive cochlea[1].

About 1 in 1,000 infants is born profoundly deaf. About half of these cases have a genetic cause. Mutations in many genes are involved — the most common, accounting for about one in five deaf children, are those affecting connexin 26.

Still, most children born to deaf couples can hear. Many of the mutations involved are recessive, which means that a baby will be deaf only if it inherits two copies of the same mutated gene. For John and Karen, the laws of inheritance could not give them a deaf child — their deafness is due to recessive mutations in different genes.

The couple's genetic counsellor is now investigating attitudes to genetic testing among the deaf. In a pilot study conducted at Gallaudet University in Washington DC, a college for the deaf and hard-of-hearing, Pandya and her colleagues asked students whether they would be interested in considering genetic test results to help them select a partner[2]. More than half of the 64 respondents said they would — but it wasn't clear from the wording of the questionnaire whether this was because they wanted a deaf child, or a hearing one. Pandya is planning a larger study to explore the issue further.

Using genetic tests to identify a partner with whom to try and have deaf children is one thing; aborting a fetus if it turns out to be able to hear is another. Evidence that a small minority of deaf people would consider this option comes from the work of Anna Middleton, a genetic counsellor at Addenbrooke's Hospital in Cambridge, UK.

Middleton's first survey was conducted at the Deaf Nation conference, a gathering of the culturally Deaf held in Preston in northwest England in 1997. Of the 87 delegates who completed the questionnaire, 14 said they would be interested in prenatal testing for deafness. Four of these said that they would prefer to have deaf children[3].

Critics argued that Middleton's study was too small, and was based on a group of Deaf activists[4]. So she polled a larger sample of the hard-of-hearing, hearing people with deaf family members, and profoundly deaf people — two-thirds of whom were not culturally Deaf. Across the deaf group, about one in five said they would consider prenatal genetic testing, mostly to prepare for the birth of a hearing or a deaf child[5].

Few of the deaf respondents said they would consider abortion, and in most of those cases, their choice was actually for a hearing child. None of those who said they would abort a deaf fetus was culturally Deaf. But three deaf people said they would consider aborting a fetus if it could hear. Two of these were culturally Deaf.

Tough choices

Middleton says that it's still unclear what people would do when faced with the choice for real. "Attitudes do not necessarily predict behaviour," she cautions. And even among Deaf activists, it's hard to find someone who will be quoted as saying they would abort a hearing fetus, because of the opprobrium they would attract. "Deaf people know that it's a very risky thing to say in public that you would consider genetic testing to have a deaf child," says Padden.

The wisdom of keeping quiet was reinforced by the controversy that engulfed Sharon Duchesneau and Candace McCullough in April 2002. A Deaf lesbian couple from Bethesda, Maryland, Duchesneau and McCullough told the *Washington Post Magazine* that they had conceived a child using sperm donated by a deaf male friend, because they wanted a deaf baby. They didn't employ genetic testing to guarantee success, but their son, Gauvin, was born deaf. While the initial article was sympathetic, many of those that followed were not. The Fox News website, for instance, ran a hostile piece, headlined "Victims from birth: engineering defects in helpless children crosses the line".

Deaf couples wanting to be sure of having a deaf child have two options. They could use prenatal genetic testing, and abort the fetus if it can hear. Or they could consider *in vitro* fertilization (IVF) combined with preimplantation genetic diagnosis to select deaf embryos for transfer to the womb. In December 2002, Monash IVF, a clinic in Melbourne, Australia, conducted preimplantation tests for a couple who wanted to exclude the one-in-four chance that they would have a deaf baby.

The Infertility Treatment Authority for the state of Victoria, which sanctioned the Monash procedure, says it would not allow a couple hoping for a deaf child to use the test. "Our policy states that the procedure should be used to avoid a genetic abnormality," says Helen Szoke, the authority's chief executive. Few other regulatory bodies have yet devised

AFP/GETTY

Handmade: the Café Signes in Paris is designed to bring locals and the deaf community together.

GAMMA

Signing is an intimate form of communication in some families.

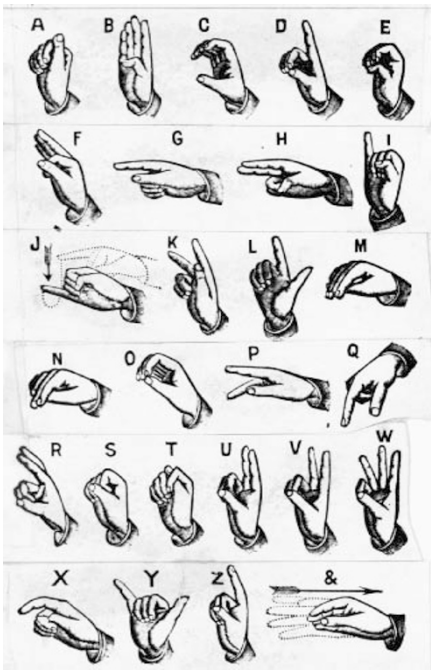
explicit policies on the issue. Britain's Human Fertilisation and Embryology Authority, for instance, which issues licences for preimplantation genetic testing on a case-by-case basis, has not yet had to rule on the matter. The UK Human Genetics Commission, meanwhile, is currently preparing a report for the government on genetics and reproductive decision-making, which may touch upon the issue.

Prenatal genetic testing for hereditary conditions is used more widely than preimplantation diagnosis. And in many countries, including the United States, there are no legal restrictions on its use. Instead, clinical geneticists and genetic counsellors would have to decide whether to assist a deaf couple to have a deaf child by giving them a test that could lead the parents to abort a hearing fetus.

An international survey of 2,906 geneticists in 36 nations revealed varying views on this point. In Norway, none of those surveyed would perform such a test, and in France, the figure was just 1%. But in the United States, Italy, Russia, Cuba and Israel, more than a third said they would[6].

In practice, such tests are far more likely to be used by hearing couples to avoid having a deaf baby. In July, *The New York Times* highlighted the case of a couple who had taken a series of genetic tests before conceiving to be sure that they weren't at risk of passing on a genetic disease. When their deaf son was born, the parents were angry that they hadn't been tested for the common mutations that can cause deafness.

BETTMANN/CORBIS



Signs of the times: an early alphabet for the deaf.

For many people born deaf, including Padden, the attitudes revealed in the piece struck close to home. "That article sent chills down my spine," she says. Middleton's surveys suggest that many deaf people feel similarly. The culturally Deaf, in particular, feel threatened by the possibility of genetic diagnosis leading to the abortion of deaf fetuses[3]. Some postings on deaf online forums have equated genetic testing with Nazi-style eugenics. Similar attitudes underpin widespread Deaf opposition to the idea of 'curing' deaf people using cochlear implants.

Testing times

This unease may explain why Middleton's surveys have shown that deaf people are less likely than the hearing to consider prenatal testing for deafness[3,5]. And among those who would consider testing, opinions vary widely. Many deaf people, for instance, are appalled by the idea of aborting a fetus if it can hear. Opinions may depend in part on whether the individual was born deaf or lost their hearing later on, and whether they grew up in a deaf family.

Given these diverse viewpoints, some experts argue that it's unfair to focus on the minority of the culturally Deaf who say they would consider aborting a hearing fetus. "It is offensive to keep harping on about this scenario. While many deaf parents may harbour a preference for having deaf children, the data suggest that the majority would never consider doing it," says Barbara Biesecker, a genetic counsellor at the National Human Genome Research Institute in Bethesda.

But if genetic testing to screen against deafness takes off, and the Deaf feel that their culture is threatened, it's possible that some will want to fight back. In this case, their best option might be to adopt the very technology they fear, and embrace genetic testing to ensure that they have deaf children.

It's even possible that some may have already done so, without anyone realizing. In many countries, there are no legal obstacles to stop a woman obtaining a prenatal test for deafness, without revealing her true motivations, and then seeking an abortion from a different healthcare provider if the result showed that she was carrying a hearing fetus. "If the question is whether there are any restraints to prevent somebody from doing this, the answer is no," says Biesecker. ■

Carina Dennis is *Nature*'s Australasian correspondent.

1. Nance, W. E. & Kearsey, M. J. *Am. J. Hum. Genet.* **74,** 1081–1087 (2004).
2. Taneya, P. R., Pandya, A., Foley, D. L., Nicely, L. V. & Arnos, K. S. *Am. J. Med. Genet.* **130A,** 17–21 (2004).
3. Middleton, A., Hewison, J. & Mueller, R. F. *Am. J. Hum. Genet.* **63,** 1175–1189 (1998).
4. Michie, S. & Marteau, T. M. *Am. J. Hum. Genet.* **65,** 1204–1208 (1999).
5. Middleton, A., Hewison, J. & Mueller, R. *J. Genet. Couns.* **10,** 121–131 (2001).
6. Wertz, D. C., Fletcher, J. C., Nippert, I., Wolff, G. & Ayme, S. *Am. J. Bioeth.* **2,** W21 (2002).