

Do children have a right to know about a parent's serious hereditary disease?

In 2007, a middle-aged British man shot and killed his wife. He was declared mentally incapacitated, convicted of manslaughter and sentenced to hospital care. Two years later, he was diagnosed with Huntington's disease – an inherited condition that damages nerve cells in the brain, causing both mental and physical deterioration until finally proving fatal – which may have contributed to his diminished mental state at the time of the shooting.

The man made it clear to his doctors and social workers that he did not want his daughters to know about his Huntington's diagnosis. He said he did not want to distress them further after the trauma he had caused by killing their mother. But around the time of the diagnosis, one of his daughters became pregnant.

Huntington's is a hereditary disease that is autosomal dominant – if one of your biological parents has the faulty gene, then you have a 50 per cent chance of carrying it as well. And if you have the gene, then you will almost certainly get Huntington's. A diagnosis can have devastating consequences beyond the initial patient.

Several times doctors discussed with the man the idea of telling his children, who were attending family therapy at an NHS-run facility with members of their father's medical team. But the man insisted his daughters be kept in the dark. "Especially the pregnant one until she gives birth," he told a social worker. He said he was afraid they might get upset, kill themselves or have an abortion.

The pregnant daughter – who we'll call Jane* – gave birth to a baby girl, unaware of her father's diagnosis and the risk that she and her newborn had of developing Huntington's. Four months later, one of her father's doctors accidentally let slip her father's condition, which had been confirmed by genetic testing months before Jane gave birth. She got tested and the results came back positive – Jane had Huntington's disease.

In a lawsuit issued in 2013, Jane claims that her father's doctors had a duty to inform her of her father's diagnosis regardless of his wishes. Had she known she had Huntington's, Jane says, she would have terminated her pregnancy rather than have a child who was at high risk of both having a life-destroying disease and being raised by a terminally ill single parent.

A lower court threw out the case. But in May 2017, the UK Court of Appeal overturned the decision. Jane's claims, it said, are arguable. That decision may have set an important precedent – that genetic testing extends the duty of a healthcare professional beyond the patient. But extending that duty also risks damaging the underlying trust between doctor and patient.

If, as expected, the case goes to trial in 2019, the court will have to decide whether Jane was owed a "duty of care" by her father's doctors even though she had never been their patient or at any of the facilities at which her father was treated. More broadly, the court will ask if the genetically inheritable characteristic of a condition like Huntington's disease means that a parent's confidential diagnosis belongs to their children as well. With further advances in genetic testing, claims to a relative's genetic information

may become increasingly difficult to deny. As one scholar has put it, it will be more common for the relative of a patient to demand access to the patient's record by declaring – with more than a hint of truth – “I have a claim, because it is about me.”

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Over the past 30 years, Heidi Pate has had seven surgeries on her neck. During the first two alone she had 108 lymph nodes removed. She is constantly trying to keep ahead of her cancer and so has allowed surgeons to open her up and move or remove everything that they think could kill her. When they took out her parotid salivary gland at age 32, her face would have been “sucked in and concave” had her surgeon not replaced the gland with muscle taken from the left side of her neck. “I guess my doctor felt sorry for me,” she tells me when I visit her home in Orange Park, Florida.

Heidi's battle with cancer didn't begin with her own condition. It began with her mother, Marianne New, who was treated for medullary thyroid carcinoma, known as MTC, in 1987. Marianne had a couple surgeries and recovered fully. But three years later, her daughter started having symptoms of what doctors mistakenly thought was stomach cancer. Just before Heidi's gallbladder was removed, an intern found swollen lymph nodes in the right side of her neck. Eight weeks later her surgeon did a biopsy and, when the results came back, said: “Yep, that's thyroid cancer.”

When you have MTC, the C cells of your thyroid gland produce an excessive amount of a hormone called calcitonin. A typical level in a person's body is less than 10 picograms per millilitre (a picogram is one trillionth of a gram). The day that Heidi had her first thyroid surgery – just a few days after the biopsy – her calcitonin level was 10,000. Actually, the doctors told her they didn't really know what her number was.

That was just the top of their scale.

As soon as Heidi was diagnosed, the doctors asked about her family. She told them she had a sister. They told Heidi to make sure that her sister got tested immediately – MTC is inheritable, and since their mother had it, there was a 50 per cent chance her sister would have it too (she tested negative). “That’s what cued me,” says Jim Pate, Heidi’s husband since they were teenagers. “Why wasn’t your mom’s doctor so concerned about you and your sister at the time of her surgery? We didn’t get a word from them.”

Jim and Heidi went to a local library to look up MTC and found a book called *Understanding Cancer*. She pulls out a packet of photocopies of the book that she still keeps today. She shows me the very first page of the chapter on thyroid cancer, where it states that MTC is often hereditary. “The *first page!*” she says. They found additional articles that showed that it had been known for years that MTC could be hereditary.

After much discussion, not least with their minister, the Pates decided to sue their mother’s physician, Dr James Threlkel. The complaint alleged that Threlkel should have known that MTC was inheritable and that he had a duty to warn Heidi’s mother that her children were at risk. If he had warned his patient, then Heidi could have started treating the cancer three years earlier, when she was asymptomatic, and potentially have been cured.

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The trial court for Alachua County dismissed their complaint. Heidi was not Threlkel’s patient, it said, and he had no duty to her. A court of appeal agreed with the previous court’s dismissal, but added that if the Pates chose to take their case up one level – to the Supreme Court of Florida – they would be asking the judges a very important question: does a physician owe a duty of care to the children of a patient – or at least have a duty to warn the patient of the genetically transferrable nature of the condition for which they’re being treated and the risk that it poses to their offspring?

The Pates did take it to the next level – and six of the seven judges of the Supreme Court of Florida agreed with them that a duty of care was owed. Reversing the previous court’s decision, they said Threlkel had failed to perform his duties by not warning Heidi’s mother of the inheritable qualities of her disease. Heidi would be allowed to take Threlkel to court. However, the judges added that Threlkel could have fulfilled his duty by simply warning Heidi’s mother of the risk to her children – he didn’t have to locate or warn Heidi or her sister directly.

“It’s arrived!” begins the animated title of a recent paper by Roy Gilbar, an honorary senior lecturer at Leicester Law School, and Charles Foster, a law professor at Green Templeton College, Oxford. Gilbar and Foster are excited because Jane’s case, they say, has the potential to redefine not only the responsibility a doctor has to an individual patient, but far beyond to what it means to be an individual in the first place.

Up until now the law has only looked at a “pastiche of a patient, a pastiche of a human being,” Foster tells me over the phone. But Jane’s case recognises that we are social animals rather than walled-off islands – that each of us has unbreakable connections to other people in our circles and that these connections must be considered in medical care. It adopts a more realistic approach towards the question of what a human being is, says Foster, and therefore what the duties of human beings towards other human beings should be.

“The law is finally catching up with the notion that the creatures that it deals with when it approaches medical law are wonderful, mysterious, complex, multi-layered creatures. Not the cardboard cut-outs that lots of medical ethics and law tends to assume they are,” Foster says. “The rather sclerosed way in which the law has assumed that there is one easily identifiable person, with an easily ascertainable suite of autonomy interests, never accorded with the realities of our daily lives.”

Jane’s case could force doctors to consider the nexus of relationships in which we exist and step out of their comfort zone when it comes to breaking doctor–patient confidentiality. Few cases have ever gone this far, and for good reason: confidentiality has been at the core of medical practice for thousands of years. It is seen as key in maintaining trust between a physician and patient, and has been the default position in international medical guidelines ever since the creation of the Hippocratic Oath.

Right now doctors need to think about their duty of confidentiality on the one hand and their duty to warn other people, like relatives, on the other, Gilbar tells me. “Up to this case they didn’t have to think about relatives. They only thought about the patient,” he says. “And if the patient said, ‘Look, I don’t want anybody to know about my situation,’ they simply respected that.” Jane has the potential to change all that.

Donna Safer was 10 years old when her father, Robert Batkin, died at age 45. He had been sick for years before that, and among the physicians who treated him was Dr George T Pack, one of the world’s most celebrated cancer specialists. In 1951 Pack had flown to Argentina to treat Eva Perón, the country’s famous first lady. But during his seven years as Robert Batkin’s doctor, Pack never told the family what was really killing his patient, says Donna.

As a young adult, Donna believed her father had had some form of liver cancer, because she remembered him having yellowish skin before he died. But she and her mother didn’t really know. They just assumed that’s what it was. In fact, as her father went through various treatments and surgeries, all Donna knew was that he had some sort of infection. When Donna’s mother asked the doctors if her children were at risk, she was told at least twice not to worry.

“You have to picture back in 1956,” Donna tells me during a long phone call from her home in Connecticut. “Cancer was a dirty word. It was like in the 1980s when you said you knew somebody with AIDS. It was like, ‘Don’t touch them because you might catch it.’ This is the mentality of cancer in the Fifties. And people were not informed by their doctors. Doctors didn’t tell people the way they do now. It was much more secretive.”

Throughout her father’s surgeries and treatments, the doctors told Donna’s mother that they were treating

her husband's "infection". But every year it seemed he would be back in the hospital, often for weeks at a time. By the early 1960s, he had had his colon removed and was using a colostomy bag. In January 1964, he died. "His death certificate says natural causes," Donna says. "That's unheard of at 45 years old."

In 1990, Donna, then 36 years old and engaged to a doctor named Robert, started gaining weight and feeling bloated all the time. One day, on her drive to work, she had to pull over to the side of the road because she couldn't keep her eyes open. A week or two later, she doubled over in pain, and she and Robert decided they had to figure out what the heck was going on with her body. After a walnut-sized object was found during a rectal exam, Donna got a barium enema. Afterwards, the radiologist came out and showed the results to Robert. Donna's colon was totally blocked and the diagnosis was clear.

"One of the worst, worst moments of my life," Robert says. "Now I have to tell my fiancée that she's got colon cancer." That was a Wednesday morning. By Friday Donna had had her colon removed.

The next year, at the suggestion of a retired family doctor, Donna looked at her father's old medical records and saw that a procedure he had had around 35 years earlier was nearly identical to hers. It turned out that Donna had familial adenomatous polyposis, which can be passed on from parent to child. She and Robert dug up old papers of Dr Pack's and discovered that by 1956 – eight years before Donna's dad died – it was already known that the condition was hereditary.

In 1992, Donna and Robert attempted to sue Pack's estate, aiming not to disgrace the long-deceased physician but to get money from the insurance company. They claimed that Pack knew the hereditary nature of the disease but did not warn Donna's parents that their children were at risk.

As in the Heidi Pate case, the trial court dismissed their claims because Donna was not Pack's direct patient. He had no duty towards her as a doctor, the judge said. Nor did her father's cancer pose a risk to the public at large – as would, say, a breakout of Ebola or another contagious disease. The risk was only to Donna and that was not enough for the judge.

But in 1996, the Superior Court of New Jersey reversed that decision, finding "no impediment, legal or otherwise, to recognising a physician's duty to warn those known to be at risk of avoidable harm from a genetically transmissible condition". The court acknowledged that extending the duty to warn non-patients "might lead to confusion, conflict or unfairness in many types of circumstances", but that the risk from genetic cases – which are limited to relatives – "is sufficiently narrow to serve the interests of justice".

Unlike Heidi's case, however, the New Jersey judges specifically said that warning the patient is not always sufficient to avoid risk to third parties such as relatives, and that a doctor's duty of care can extend outside the hospital even without a big picture public health risk. While the court didn't rule on how doctors might go about warning third parties who are not their patients, it did decide that a duty to them could be owed. Moreover, it predicted that "it may be necessary, at some stage, to resolve a conflict between the physician's broader duty to warn and his fidelity to an expressed preference of the patient that nothing be said to family members about the details of the disease".

Twenty-three years later, that prophecy has materialised in the UK. Jane's case could force doctors to

extend their duty even further, or at least to question to whom one patient's diagnosis belongs.

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For Graeme Laurie, a professor of medical jurisprudence at the University of Edinburgh, Jane's case represents a frightening scenario. He believes it could create uncertainty about the nature and extent of legal duties for healthcare professionals: uncertainty about confidentiality, uncertainty about to whom a doctor owes their primary responsibility – uncertainty that fundamentally changes the relationship between medical professionals and their patients. "It's not necessarily always clear to me," says Laurie, "that the so-called public interest in disclosing the information would be strong enough in future cases to justify such a breach."

In the very extreme and tragic circumstances of Jane's case, it might make sense to argue a duty of care, he says. But imposing a legal duty on doctors to disclose patient information in broader circumstances may cause them to do so for legal reasons rather than medical ones.

"With a legal duty it can too easily become a knee-jerk reaction; I have to disclose, come what may, because I now fear liability. A fear of liability is not acting in the interests of anybody. A fear of legal liability is the fear of being sued or fear of getting sacked because your employer will have to pay money. It's not about an ethic of care," says Laurie. "My concern is replacing a discretion with a duty."

Doctors in the UK already have a good deal of discretion when it comes to breaching confidentiality. In fact, Laurie believes that Jane's father's doctors would have had a sound argument, based on current standards, for breaking with his wishes. A 2011 guideline from the Royal College of Physicians, the Royal

College of Pathologists and the British Society for Genetic Medicine offers clear leeway: “Where consent to release information has been refused... the rule of confidentiality is not absolute,” it states.

“You don’t have to keep all confidences irrespective of circumstances. Of course not. Somebody confesses a murder, you’re entitled as the healthcare professional to go to the police,” says Laurie. Jane’s case could have been an exception as well. “They could have approached the daughter and told her the circumstances, even despite the fact that the gentleman in question was objecting.”

Perhaps the doctors could have told Jane that she should get tested for Huntington’s, allowing her to infer on her own that her father had the disease. “There are circumstances where we are allowed to breach confidentiality and this is one of them,” says Anneke Lucassen, a clinician and professor of clinical genetics at the University of Southampton, who is serving as an expert for Jane’s legal team. “They could have done that under existing law.”

Lucassen is an author of a recent paper exploring the attitudes of healthcare professionals, patients and the public regarding disclosure of patient information. The results, drawn from 17 different studies, found that most patients tell, or intend to tell, at-risk relatives of genetic or inheritable conditions. But some do not for reasons including “guilt and fearing blame, poor relationships, difficulty finding the right time, and perceived inability to communicate accurately”.

In one study, 30 per cent of patients said that they would not mind if healthcare professionals gave out their medical information to relatives without permission, but 50 per cent thought they should be punished for doing so. In another study presenting three scenarios – Huntington’s disease, breast cancer and colon cancer – less than one-quarter of surveyed respondents thought healthcare professionals should “seek out and inform relatives about risk and prevention options against patients’ wishes”. In a study that asked participants to take the relatives’ perspective, more than half of those surveyed said they would want a healthcare professional to disclose against the patient’s wishes if the condition were nonfatal and treatable (unlike Huntington’s) with a 50 per cent chance of showing symptoms of the condition.

One overall takeaway of Lucassen’s review is that “participants generally thought privacy was less important than the opportunity to prevent serious illness or death”. However, a somewhat conflicting conclusion found that patients believe “that unsolicited contact with relatives could invade their privacy”.

Then there’s the question of logistics. If doctors all of a sudden are responsible to the relatives of patients, are they obligated to track them down wherever they reside or else be liable? “I think most clinicians would say they do not want that, and I would agree,” says Lucassen. “It doesn’t make sense to give clinicians a duty of care to relatives because it’s just not a duty that could possibly be enforced consistently and thoroughly.” But, she adds, that’s not the same as saying you never tell anyone anything.

And what about the right *not to know*? What about those family members who don’t want to know that they have a horrible disease such as Huntington’s? Some people may be damaged by information given to them by a healthcare professional they didn’t even employ. As Laurie says, “Not only am I going to experience an early and horrible death, but now I’m doomed to live with the knowledge of that and I can’t un-know something that you told me. And you’re telling me that because you’ve been legally obliged to do

so.”

When it comes to genetic tests, two international conventions – the UNESCO International Declaration on Human Genetic Data and the Oviedo Convention in Europe – recognise the right not to know results. If doctors are legally obliged to tell third parties about genetically inheritable diseases they are carrying, we could all lose what seems a reasonable right to enjoy our lives in ignorant bliss.

Though the New Jersey court agreed that Donna Safer should be able to argue her case, when it went back to trial, she lost. She tells me she had the opportunity to settle, and regrets not taking it on advice from her lawyer. However, she’s proud that her case is studied in law schools and dissected in academic articles. “I would say in the long run, I won,” says Donna, who went through another major surgery in May 2018. “If I can save one person, then I’ve made a major accomplishment.”

Rather than go to trial, Heidi and Jim Pate went to mediation with Dr Threlkel and eventually settled. Heidi says that at one point during the discussions, Threlkel’s side implied that perhaps Heidi’s mother had been informed about the hereditary nature of her cancer after all, but neglected to tell her daughters. Heidi quashed that idea, describing how responsible and caring her mother was and remains to this day. “She would have told us immediately,” Heidi says. Besides, Jim adds, “What mother would not relay that information to one of her only two daughters?”

Both of the Pates’s children tested positive for MTC and had had their thyroids removed by the time they turned five years old. Neither has shown signs of cancer since. Heidi wishes she had had the same opportunity. She believes she could have been freed of cancer if it hadn’t spread to her lymph nodes, which may have occurred during the three-year gap between her mother’s diagnosis and her own. Her calcitonin level is now back in the 5,000-picogram range, and doctors search her body every six months in an attempt to find out where the cancer is growing this time.

I told the Pates about Jane’s case and the worry that it could theoretically push the duty of care too far in the UK. They said that their lawyer mentioned a similar concern with Heidi’s case back in the early 1990s, when HIV was still essentially untreatable and killing thousands of Americans. The lawyer said the judges would not want to set a precedent that if you have HIV, your doctor would have to tell every person you’ve ever had sex with. “They wanted to put some sort of a dam up to hold that back, because they didn’t want to turn this into a free-for-all,” Heidi tells me. But in the end, the Florida court’s decision to allow Heidi to argue her case did no such thing. It did not open any floodgates.

Jane’s lawyer, Jonathan Zimmern, believes that clinical geneticists are already practising medicine in the manner that Jane wishes her father’s doctors had. He’s spoken, informally, with many clinical geneticists about the case. “They all feel that what we were fighting for is what clinical geneticists do on a daily basis,” he tells me.

“[Clinicians] already act and feel as though they have a professional duty to someone other than their patient or the person who provided the original genetic sample,” Zimmern says. And if they already practise this way, he doesn’t understand why the NHS is so adamant about denying Jane in court. “It seems to me they are fighting a battle on behalf of doctors who don’t want the battle to be fought.”

That said, Jane could alter the law by legally extending a doctor’s obligations. “It’s a very minor extension of the duty that clinicians were already operating under,” says Zimmern, “but it is kind of a significant development in the law, just because we’ve always had that line in the sand – that the clinician’s duty is only to the patient and to no one else.”

Zimmern says that the UK Court of Appeal has, in a sense, already pushed past a new boundary. An important legal precedent has already been set: that arguably in genetic cases there is a duty of care to someone other than the patient. “If we win the underlying case now, it’s kind of just reinforcing what is already there. But if we lose, it doesn’t overturn the Court of Appeal decision,” says Zimmern. “As the years or the months go by, if any other case arises like this, everyone is going to rely on that decision, and it’s likely to become law.”

A common saying among legal practitioners is that hard cases make bad law. “My concern as a lawyer is I don’t really trust the law,” says Graeme Laurie. “I don’t trust the law not to run away with the precedent and introduce more uncertainty into the doctor–patient relationship.”

Whether Jane will, or should, open the floodgates is a difficult question. Any precedent that will change the dynamic of doctor and patient must take into account a nearly infinite number of complexities. Perhaps it’s better to keep things simpler, to maintain that doctors are responsible to their patients only, except in truly extraordinary circumstances. “It’s a genuine dilemma,” Laurie says. “It’s not absolutely clear what is the right thing to do.”

**Jane – known as ABC in the lawsuit – has been kept anonymous because of privacy concerns for her young child.*

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