

Education and debate

Ethics in practice

Revealing the diagnosis of androgen insensitivity syndrome in adulthood

Jennifer Conn, Lynn Gillam, Gerard S Conway

It is always going to be difficult for a woman to find out that she is genetically male. What are the ethical issues generated by being confronted by outdated practice on disclosure?

This article is part of an occasional series, edited by Michael Parker and Julian Savulescu, analysing ethical issues that confront health professionals in daily practice

Complete androgen insensitivity syndrome, previously called testicular feminisation,¹ is a rare X linked condition with an incidence of between 1:13 158 and 1:64 200 live births.²⁻⁴ An affected woman has a 46XY karyotype that leads to normal differentiation of testes in utero, but a defect in the gene coding for the androgen receptor results in complete insensitivity to circulating androgens, resulting in phenotypic female development.⁵ Psychosexual orientation is in every respect female. There is, however, no uterus and only a partially formed vagina, and pubic and axillary hair is scant or absent.¹⁻⁵

Case history

A gynaecologist who was retiring from clinical practice referred a 40 year old woman with complete androgen insensitivity syndrome to an endocrinology clinic for ongoing follow-up. The patient's records stated she had had hernia surgery in infancy, probably the removal of a testis. At 17, she had presented with primary amenorrhoea, and investigations showed she had complete androgen insensitivity syndrome. Neither she nor her family were told the underlying diagnosis, as was common practice at the time. They were informed that she had been born without a uterus and that she had only one "ovary," which had malignant potential. She proceeded to gonadectomy and vaginoplasty and started oral oestrogen replacement.

She had been devastated to discover that she would be unable to bear children and intermittently attended for counselling with a clinical psychologist who was aware of the underlying genotype. She subsequently developed a satisfying long term sexual relationship, but her partner died unexpectedly, and to deal with her grief she returned to the psychologist.

She told the psychologist that she despaired of finding another partner who would be so understanding of her infertility and expressed confusion about why she had been born without a uterus. She, nevertheless, was acutely aware of her physical attractiveness and was recorded as saying: "At least I can take comfort in my femininity." The psychologist had written in the medical notes that because of her



fragile emotional state she, "should never be told the true diagnosis."

Concealment of the diagnosis is now considered outdated. The doctor who first saw the patient in the clinic was placed in a difficult situation. Should he rigorously adhere to current best practice or somehow take into account her personal history and the established approach at the time of diagnosis? To complicate matters, she was leaving the next day for three months in South East Asia, leaving no immediate opportunity to tackle the problem. He arranged a review appointment and set about exploring the underlying historical and ethical issues to construct an optimal plan of management.

Historical context

In a seminal paper published in 1953, the American gynaecologist John Morris described the anatomical, histological, and clinical features of androgen insensitivity syndrome, based on 82 cases collated from over nearly 150 years of the medical literature.¹ He argued against disclosure of genotype: "It goes without saying

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that it would be unwise to inform the patient of the true state of affairs ... it seems only necessary to state that childbearing is impossible."¹ He expressed concern about the apparent high prevalence of psychiatric morbidity among people with intersex conditions, suggesting that this often resulted from "overzealous medical attention." He wrote compassionately: "The obvious humane attitude is not to interfere and, by such, [avoid] meddling [that might] produce a psychiatric casualty and perhaps suicide."¹

Morris's paper arguably established the tenor of the psychosocial management of complete androgen insensitivity syndrome for decades. His approach to non-disclosure was consonant with the prevailing medicolegal and ethical climate of the 1950s. Under the then widely accepted ethicolegal principle of "therapeutic privilege," it was legitimate for a doctor to withhold information from a patient if revealing that information was thought not to be in the patient's best interests.^{6,7} Therapeutic privilege was still widely applied at the time of our patient's initial presentation as an infant in the 1960s. Acceptance of this principle has, however, fallen steadily over time, in line with the increasing importance given to informed consent and autonomy,^{6,7} and it is now considered to have limited usefulness.

The specific issue of revealing the genotype in complete androgen insensitivity syndrome at diagnosis remained contentious well into the 1990s.⁸⁻¹³ Minogue and Taraszewski argued that the diagnosis could justifiably be withheld if the clinician thought the patient or her family could not deal with the revelation.⁸ Shah, writing in 1992, argued that: "The disclosure of genotype is irrelevant to care and may be confusing to patient and family."⁹ As late as 1997, in a major biomedical review of the molecular biology of the androgen receptor and its role in sexual differentiation, Weiner et al wrote that the question of whether to tell a patient about her genetic make-up was complex and acknowledged the disparate views expressed in the literature.⁵

It is now established practice to disclose the genotype of complete androgen insensitivity syndrome at diagnosis. Nevertheless, a cohort of women whose management was founded in a different social and ethicolegal context remain unaware of their biological make-up well into adult life. Some of these women have been lost to follow up, but in other cases, clinicians will have made a considered decision to maintain the silence, perhaps because of anxiety about causing harm or a perceived difficulty in broaching such a sensitive issue. In this case, the patient's previous healthcare providers had decided that she was psychologically too fragile to receive the diagnosis. To explore how her new clinician should incorporate this information into her clinical management we turned to the commonly applied four principles approach of Beauchamp and Childress.^{6,7}

Applying normative ethical principles

Beneficence

The ethical principle of beneficence obliges medical practitioners to treat their patient in a way that produces maximum benefit for that person.⁶ On one hand, our patient's interests would arguably best be

promoted if she had a full understanding of her condition, particularly since she has expressed confusion about the reason for her absent uterus. Knowing her diagnosis may also provide options that optimise her welfare—for example, she could join a support group and meet women with the same condition.¹²

On the other hand, she is known to highly value her femininity, and her self image as a female may be challenged by knowledge of her genotype, no matter how skilfully this information is communicated. It is difficult, however, to decide what is best for the patient without a full understanding of her and her world; indeed, it would be parentalistic to attempt to do so on her behalf.

Non-maleficence

The duty to avoid causing harm,⁶ or non-maleficence, was no doubt foremost in the minds of previous healthcare providers when they adopted a policy of non-disclosure. Telling the patient would, at least, cause her unease; at worst, it could be disastrous. The psychologist had already hinted at the potential risks. When the case was discussed at our clinic meeting, a visiting professor reported a similar situation; the patient had become deeply troubled when informed of her genotype and committed suicide.

What harm might come from continuing to conceal the diagnosis? One possibility is that she might find out the truth by chance. Dreger reports on a woman who learnt her diagnosis, alone and frightened, in a medical library; alienated from her healthcare providers, she avoided medical care for 18 years with the result that she developed severe osteoporosis.¹⁰ This is not an isolated case.¹⁰ Advances in information technology have exponentially increased the chance of a woman finding out her diagnosis. When we entered "absent uterus" into the internet search engine Google, it retrieved 104 000 websites, with many of the first listed sites referring to androgen insensitivity syndrome. In addition, there were over 30 000 websites specifically dedicated to this genetic disorder.

Other circumstances might lead our patient to discover her condition. A new sexual partner might question her surgical scars and lack of pubic and axillary hair; a health professional with access to her medical file might unintentionally or deliberately, yet insensitively, reveal the diagnosis. Or she may simply become more curious about her medical history and request access to her file. Each of these scenarios has arisen for patients now attending the intersex clinic at the Middlesex Hospital.

If a policy of non-disclosure is maintained, she might never discover the truth about her genetic identity but continue to have unease about why she does not have a uterus. Most patients can sense when a secret is being kept.¹⁴ Dreger has cautioned: "Hiding the facts doesn't stop a patient ... from thinking about it ... indeed the failure on the part of the doctor ... to talk honestly about the condition is likely only to add to feelings of shame and confusion."¹⁰

Autonomy

Whereas beneficence and non-maleficence are mostly considered in terms of consequences, the principle of autonomy focuses on rights and obligations.⁶ It asserts the rights of patients to make decisions about their medical treatment, "Free from controlling influences

or personal limitations such as inadequate understanding.¹⁶ It confers an obligation on healthcare providers to be truthful and to respect the choices that their patients make.

Our patient currently does not need to make any decisions about her physical treatment. Her gonads have been removed, and she is no longer at risk of malignancy. She has been taking oestrogen daily, and her bone density is within normal limits. Nevertheless, knowing about her genetic make-up might enable her to exercise choices in other aspects of her life. She could, for example, choose to share information about this X linked condition with other members of her family.

An argument could be made for non-disclosure if she were not able to function autonomously. If she was intellectually disabled or had an acute psychiatric condition, she would have limited capacity for information processing and self determination. The clinical psychologist described her as being too fragile to be told about her genotype, but there does not seem to be any evidence that this is still the case.

Respect for patient autonomy would also allow the diagnosis to be withheld if the patient did not want to know. But she does not know that there is something to disclose. Unless we tell her, we cannot find out whether she wants to know. We have no evidence that she does not want to find out about her underlying diagnosis—the fact that she has expressed confusion about her absent uterus indicates that the reverse is likely to be true.

Justice

Justice is in its broadest sense about equality and fairness.¹⁵ It raises several issues pertinent to this case. In terms of equality, it suffices to apply Rawls' strategy of asking what we would want to happen if the positions were reversed.⁶ Our patient deserves respect and it is only fair that she be told her genotype similar to anyone else in her situation; the sensitive nature of her condition does not abrogate the obligation to disclose. In addition, it is only fair that she is given access to the clinical expertise of her healthcare providers who can inform her sensitively. In terms of distributive justice, sufficient resources are available; other patients will not be deprived of medical care if time is allocated to her.

Decision

Any obligation to respect the decisions of the previous healthcare providers is clearly over-riden by the ethical principles of justice and respect for patient autonomy. The argument that disclosure would lead to unacceptable harm is no longer valid. Unquestionably, the great-

est harm would result if the patient found out her diagnosis by chance in an unsupported environment. The probability of this happening through modern information technology is high. The harm associated with disclosure, conversely, is likely to be short term and minimised by a sensitive and skilful approach.

When the patient returned to clinic, the doctor asked whether she had ever been curious to know why she did not have a uterus. She said yes, quickly adding that she was sure that information had been withheld in the past. He arranged for her to see a clinician with expertise in dealing with intersex conditions. She was initially extremely angry when told about her genotype, but knowing about it has led to a positive outcome. She continues to attend for regular follow up.

The series is edited by Michael Parker, reader in medical ethics at the Ethox Centre, University of Oxford (michael.parker@ethox.ox.ac.uk) and Julian Savulescu of the Oxford Uehiro Centre for Practical Ethics. We thank the patient for supporting the writing of the paper and Jeanette Lawrence for her helpful comments.

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War wound

About 10 years ago I was asked to see a man in his 90s with symptoms of "piles." He was relatively fit and mentally alert, so he was able to give me the longest and most accurate history of onset that I have ever had.

He told me his problems had begun with constipation, which he had while serving with his regiment at the battle of the Somme in 1916—the trench latrines had been destroyed by shellfire. In 1919, after he had been discharged from the army, he

consulted a doctor, who advised him that surgery could not be recommended and that he should continue to use the cream. He did so for more than 70 years until he went into care. Fortunately, a couple of elastic bands relieved his symptoms.

Could his chronic long term disability be described as a battle injury?

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