

Sex and gender issues in competitive sports: investigation of a historical case leads to a new viewpoint

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ABSTRACT

Based on DNA analysis of a historical case, the authors describe how a female athlete can be unknowingly confronted with the consequences of a disorder of sex development resulting in hyperandrogenism emerging early in her sports career. In such a situation, it is harmful and confusing to question sex and gender. Exposure to either a low or high level of endogenous testosterone from puberty is a decisive factor with respect to sexual dimorphism of physical performance. Yet, measurement of testosterone is not the means by which questions of an athlete's eligibility to compete with either women or men are resolved. The authors discuss that it might be justifiable to use the circulating testosterone level as an endocrinological parameter, to try to arrive at an objective criterion in evaluating what separates women and men in sports competitions, which could prevent the initiation of complicated, lengthy and damaging sex and gender verification procedures.

INTRODUCTION

In 1949, the Dutch track athlete Foekje Dillema (1926–2007) came to prominence on the world athletic stage. She started to rival Fanny Blankers-Koen, the world-famous Dutch track athlete who won four gold medals during the 1948 Summer Olympics in London and was elected Female Athlete of the Century by the International Association of Athletics Federations (IAAF) in 1999. In contrast, Dillema's career was of short duration, with a dramatic ending. In 1950, she was expelled for life by the Royal Dutch Athletics Federation, due to the results of a 'sex test', for which details or results were never revealed and no records are available. Her 1950 national record of 24.1 s for the 200 m, which she took from Fanny Blankers-Koen, was erased, and only after her death 57 years later was she reinstated by the Royal Dutch Athletics Federation (figure 1).¹

The verification of the sex of athletes has been an issue for many decades. It should be noted that reports and reviews on this topic refer to gender verification, rather than sex verification.^{2–4} However, what counts in competitive sports is a person's sex characteristics. Trying to avoid the word sex, given its charged nature, can only cause confusion.^{5,6} Herein, we will use the term sex for the biological and physiological characteristics that define men and women, as compared to gender and gender identity in reference

to the socially and individually perceived sexual identity of an individual from birth to puberty and adulthood.^{5,7,8}

Sex verification in 1950 was based solely on physical examination, predating hormone assays or sex chromosome analysis. Following discovery of the Barr body in female cells in 1949, it took some 12 years before it was known that this body represents an inactivated X chromosome⁹; from the late 1960s its detection was used in sex verification tests in the context of sports competitions.^{2–4} Subsequent tests focused on the male-specific region of the Y chromosome, particularly the male sex determining *SRY* gene.¹⁰ However, opposition to sex verification for female athletes with laboratory-based genetic testing developed in the 1970s and 1980s, because these tests did not encompass the complexities of disorders of sex development (DSDs). Since the 2000 Summer Olympics, questioned sex and gender is evaluated on a case-by-case basis by a team of specialists in the areas of endocrinology, genetics, gynaecology and psychology.^{3–5}

To broaden the perspective on sportswomen confronted with questioned sex characteristics, we have investigated the case of Foekje Dillema, with informed consent from her heirs, by means of DNA analysis of samples from worn clothing. Appreciating the nature of the samples tested, we applied DNA methodology and lab quality standards used in human forensics. Our DNA analysis indicates that Foekje Dillema had a 46,XX/46,XY mosaic condition with a rare origin, which we interpret as leading to hyperandrogenism from her puberty. Based on this historical case we discuss that, if a sportswoman is confronted with signs of a DSD early in her sports career, it is harmful and confusing to question such a person's sex and gender. Rather, we suggest that it is necessary to try to arrive at an objective criterion in evaluating what separates women and men in sports competitions.

RECONSTRUCTION OF A HISTORICAL CASE

From the combined genotyping and DNA quantification results, we conclude that Foekje Dillema was a 46,XX/46,XY mosaic, with equal numbers of both genetic cell types at least in her skin (online supplementary data). In the fetal gonads of a 46,XX/46,XY mosaic, the tissue ratio of XX:XY cells will push the bipotential gonads to become either ovaries or testes, or both. A preponderance of 46,XX cells in fetal gonads can



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Figure 1 Foekje Dillema (in white shirt on the left) together with Fanny Blankers-Koen, on the Olympic Day, 18 June 1950, in the Olympic Stadium, Amsterdam, when 60 000 spectators witnessed Dillema winning the 200 m in 24.1 s, in a race in which Blankers-Koen did not participate.¹ Photo: Ben van Meerendonk (collection International Institute of Social History, Amsterdam).

lead to the development of ovaries, but with some 46,XY testis tissue present in one or both of these ovaries. Such an ovotestis condition, which can also occur in the form of a complete ovary and a complete testis on either side, has been referred to with the term true hermaphroditism.^{11 12} The most common karyotype found in true hermaphroditism is 46,XX, followed by 46,XX/46,XY chimerism and mosaicism.¹² We would like to emphasise that the term hermaphrodite, as well as other terms such as intersex, need to be replaced by the DSDs classification proposed by consensus in 2006,¹³ for the simple reason that the terms hermaphrodite and intersex cannot, and should not, be applied to human individuals. Instead, a true hermaphrodite is correctly referred to as either a female or male individual with ovotesticular DSD.¹³ When ovotestis formation is confined to only one of the gonads, the other gonad and the ovarian part of the ovotestis can function as steroidogenic ovarian tissue.^{11 12} The testicular part of the fetal ovotestis will produce anti-Müllerian hormone, insulin-like factor 3 and testosterone,¹⁴ acting towards regression of Müllerian ducts, initiation of testis descent and partial virilisation by circulating androgens, respectively, but none of these effects might reach a level where overt fetal virilisation leads to the birth of a boy. Hence, an individual with ovotesticular DSD often is raised as a girl,¹² and also experiences a female gender identity (described below).

The X:Y ratio in the adult skin, as we obtained from Dillema's clothes, does not provide any information about the ratio in her fetal gonads. However, Foekje Dillema was formally registered as a female at birth, and she was raised as a girl, according to all accounts, including those available from her family. Hence, she is unlikely to have been exposed to a markedly elevated level of testosterone during fetal development. From all available data, including the present DNA evidence, we deduce that Dillema had an ovotesticular DSD with a predominance of ovarian tissue. At the onset of puberty, gonadotropic stimulation of the gonads most likely led to marked activation of steroidogenesis – the production of both

oestradiol and testosterone in Dillema's case of ovotesticular DSD. From photographs, it is evident that from puberty she had breast development, but personal accounts also indicate that she showed some facial hair growth.¹ Hyperandrogenism from puberty may have contributed to Dillema's athletic performance.

SEX CHARACTERISTICS, GENDER IDENTITY AND TESTOSTERONE

To have, or not to have, a Y chromosome is the primary decisive factor in human sexual differentiation, but there are exceptions. A prominent example is offered by 46,XY females who have the complete form of androgen insensitivity syndrome (cAIS), when the testes produce testosterone but the body is not able to respond to androgens (testosterone and its more powerful metabolite dihydrotestosterone) due to mutation of the X-encoded androgen receptor.¹⁴ Consequently, these individuals are born and raised as girls, and have a female gender identity.^{15 16} Action of testosterone through binding to the androgen receptor in the developing fetal brain is the predominant factor in programming human male gender identity,^{7 17} and the female gender identity of 46,XY cAIS women is explained by loss of this androgenic effect. In sports, 46,XY cAIS women can be expected to have a disadvantage compared to 46,XX women with a functional androgen receptor, the latter profiting from stimulation of muscle strength by a low level of circulating testosterone.^{18 19}

The biological basis for sex segregation in sports is the consequence of long-term endogenous androgen exposure of men after puberty.²⁰ It cannot be excluded that proteins encoded by genes in the male-specific region of the Y chromosome (MSY)²¹ might act together with androgens, widening the physiological gap between women and men. However, such a role for MSY genes will be minor, compared to the predominant role of androgen action. In men, the postpubertal testosterone level is a proven dose-dependent factor when muscle strength and other physiological factors such as the blood haemoglobin level come into play.²² A moderate pubertal and postpubertal excess of testosterone in a young woman can give extra muscle development and other signs of hyperandrogenism, but it would be a rude error to even suggest that this would affect her female gender identity.

Competitive athletes exploit fortunate combinations of natural differences in physical and mental personal characteristics, including individual variation of the endogenous testosterone level. The World Anti-Doping Agency states that an athlete's sample will be found positive if the concentration of an endogenous androgenic steroid hormone is above the range normally found in humans, and is not likely consistent with normal endogenous production, unless the elevated concentration of the steroid hormone (or metabolites or markers) is attributable to a physiological or pathological condition.²³ Strictly speaking, a female athlete is free to benefit from any endogenous source of androgen production. Some female athletes may benefit, probably to a small extent, from increased androgen production originating from a polycystic ovary.¹⁹ This is viewed as acceptable by the IAAF, who stated that conditions that may provide some advantages but nevertheless are acceptable include congenital adrenal hyperplasia, androgen-producing tumours and an ovulatory androgen excess associated with a polycystic ovary.²⁴ According to these regulations, hyperandrogenism caused by ovotesticular DSD would be unacceptable only if sex and gender

What is already known on this topic

The complex biology of sex development and its disorders appears to preclude a swift and objective assessment of the eligibility of specific women athletes to compete with other women in competitive sports. Prominent cases, historical and recent, have suffered much confusion and resulted in lengthy procedures, harmful to both the respective athletes and to sports and society at large.

What this study adds

Describing a historical case, this study puts forward the notion that societal appreciation of sex and gender issues in highly competitive sports requires discussion and understanding of relevant biomedical knowledge. However, the authenticity of an adult individual's sex and gender identity should not be questioned. Rather, there is a need for an objective and relevant criterion in evaluating what separates women and men in sports competitions.

verification would provide evidence that the female athlete in fact is a man. However, we consider it highly unlikely that any individual would aim to participate in sports competitions in conflict with his or her gender identity. There is no problem in sports at large that warrants an examination, initiated by a sports federation, of the authenticity of an adult individual's sex and gender. Hence, there is a need to reconsider the situation.

It might be considered to set an upper limit for the circulating total testosterone level for sportswomen. In cases of CAIS, a high testosterone level would be of no significance. In any other case where the total testosterone level is found to exceed a set limit, causes and consequences need to be resolved before the sportswoman (re-)enters sports competition. This would be ethically justifiable, given the fact that it would be in the individual's own interest to prevent symptoms of long-term hyperandrogenism. The causes and consequences of a high-testosterone level can be dealt with in private, not in public, and, most importantly, without questioning gender identity. The eligibility of Dillema to compete might still be questioned, even today in the current era of improved knowledge about DSDs. However, if an increased endogenous testosterone level would have been detected, possible treatment to lower this level would have cleared the way to competition re-entry, leaving no trace of a sex and gender discussion.

Obviously, a proper definition of an upper limit for the endogenous testosterone level will require a detailed discussion about measurement, metabolites, circadian and other variations, binding proteins, etc.^{25–27} Normative ranges have not been well established,²⁸ but available data indicate that a circulating total testosterone level of 3–4 nmol/l normally will not be exceeded by women of younger age.^{18,29} Leaving all antidoping controls fighting against the use of exogenous androgens in place,³⁰ it might be relatively straightforward to arrive at a consensus about the maximally allowed endogenous total testosterone level. With 8–12 nmol/l total testosterone being considered as a lower limit which may require substitution in

men,²⁷ and with a reference range of 11–35 nmol/l for men,³¹ there is a substantial and significant gap in the testosterone level between women and men.

It has been argued that sex is not a binary quantity, with the far-reaching implication that sex segregation in competitive sports is an inconsistent and unjust policy.³² This argument was substantiated by pointing out that an individual's genetic background may cause a differential sensitivity to testosterone. Indeed, a genetic polymorphism such as the CAG repeat polymorphism in exon 1 of the gene encoding the androgen receptor affects the sensitivity of cells and tissues to androgens.³³ However, this effect is likely far too small³⁴ to provide any female athlete with an advantage bringing her on par with male athletes. Such a common genetic variation should not be taken into account and does not obstruct the prevailing thought that women and men are to compete separately, meaning that there is a need for a dividing line.³⁵ Thinking about a dividing line, there is much agreement that current principles and procedures need to be revisited.^{35–37}

The historical case described herein concerns ovotesticular DSD, where the amount of steroidogenic testicular tissue will determine if the affected person develops as a woman or as a man, regarding both gender and sexual characteristics. As such, this type of DSD can be viewed as a paradigm, demonstrating that the testosterone level might offer an objective parameter to separate the sexes, if required. In fact, this parameter is already implemented, in the context of sports. Athletes (46,XY and androgen sensitive) who have undergone male-to-female sex reassignment are welcome to engage in sports competitions from 2 years after the sex change, as of the Olympic Games 2004 in Athens, according to fortunate and emancipative regulations by the International Olympic Committee.³⁸ Similarly, athletes (46,XX and androgen sensitive) who have undergone female-to-male sex reassignment can compete, but they will receive exogenous testosterone. For female-to-male sex-reassigned individuals receiving testosterone supplementation, a total testosterone level of around 30 nmol/l has been reported.³⁹ Perhaps, one day we may witness a talented 46,XX sex-reassigned male who is able to successfully compete with 46,XY males, thanks also to approved testosterone supplementation. Men exposed to stress and exhaustion face difficulty in maintaining their endogenous testosterone level,^{40–42} which might imply an advantage for 46,XX sex-reassigned males, particularly in endurance sports. Similarly, a therapeutic use exemption for long-term testosterone administration in 46,XY men, to compensate for a secondary loss of gonadal testosterone production, might provide an advantage. The above serves to illustrate the point that current concepts and regulations regarding the relationship between sex and testosterone in sports offer room for consideration. We feel that this should be taken as a starting point to discuss the circulating testosterone level as a relevant criterion in evaluating what separates women and men in sports competitions.

The present report is not meant to provide a guideline, which would require detailed analysis of total testosterone levels in large numbers of female and male athletes in relation to possible confounding factors, and consensus meetings. Rather, we aim to contribute to an open discussion involving experts from the fields of biology, medicine, genetics, psychology, sports and ethics, to accomplish a procedure which would respect the authenticity of an adult individual's sex and gender identity.

Note On 12 April 2011, the International Association of Athletics Federations (IAAF) announced the adoption of new rules and regulations governing the eligibility of females with hyperandrogenism to participate in women's competition, which will come into force from 1 May 2011 (<http://www.iaaf.org/aboutiaaf/news/newsid=59746.html>).

It appears that these new IAAF rules, as announced, are in full agreement with the viewpoint expressed in our article, which at the time of the IAAF announcement was already in press with the *British Journal of Sports Medicine*. We would like to emphasise that our viewpoint was composed independently from IAAF, and that none of the authors has been in contact with the respective IAAF expert working group.

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Contributors KNB and MK designed the experiments, KNB carried out the experiments, MK provided lab equipment and test materials, all authors contributed to the data interpretation, the writing of this manuscript was led by JAG, and all authors approved the final manuscript.

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Supplementary Data

Experimental Methodology

The samples from Foekje Dillema's clothing were collected and DNA analysis was performed under conditions and with methods commonly applied in forensic laboratories.[1-3] We carried out multilocus autosomal as well as X- and Y-chromosomal short tandem repeat (STR) profiling used for forensic human identification purposes, as well as sex chromosome quantification analyses targeting several X and Y regions.

Biological materials (putatively skin cells, sebaceous oils and sweat) were recovered from three items of clothing known to belong to Dillema using the double swab method.[3]

Specific areas of the clothing that were most likely to contain only Dillema's biological material (namely the inside armpit seam, the inside back collar, and the inside seam of the sleeves) were targeted for sampling, to reduce the potential for contamination. DNA was extracted using the QIAamp DNA Micro Kit (Qiagen), in accordance with the manufacturer's recommendations, and six independent samples contained DNA of sufficient quality for further analysis. Each sample was analysed in triplicate, for each quantification and genotyping system employed.

Autosomal STR profiling was performed with the AmpF/STR Identifiler PCR Amplification kit (Applied Biosystems) and the PowerPlex 16 system (Promega) commonly used for forensic human identity testing. The two commercially available kits amplify a total of 17 autosomal loci, as well as the X-chromosomal amelogenin gene *AMELX* and the Y-

chromosomal *AMELY*. Y-chromosomal STR genotyping was performed with the AmpF/STR Yfiler PCR Amplification kit (Applied Biosystems) amplifying 17 loci scattered around the chromosome. Eight X-chromosomal STRs, encompassing 4 linkage groups, and *AMELX* and *AMELY*, were amplified with the Mentype Argus X-8 PCR Amplification kit (Biotype). Each kit was used strictly in accordance with the manufacturer's validated protocols. Peak height ratio of the *AMELX/AMELY* loci from each of the commercial kits allowed relative X:Y quantitation.

All STR profiles were interpreted in accordance with guidelines proposed for low copy templates.[1] Specifically, any allele must have been replicated in at least 3 independent amplifications to be considered genuine. This is considered standard operating practice for amplifications of less than 100pg of template DNA, and/or highly degraded DNA. Stutter alleles in the n-1 position were not called if the peak area was less than 15% of the parent peak area. Any peak below 50 RFU was not called as genuine, to prevent spurious alleles being included.

Real-time PCR quantification of autosomal (specifically the *hTERT* gene locus on 5p15.33) and Y-chromosomal (specifically the *SRY* gene locus) DNA was performed with the Quantifiler Duo DNA Quantification kit (Applied Biosystems). Relative X:Y quantitation was performed investigating the *PCHD11X* and *PCDH11Y* homologous loci, with a 90bp deletion on the X chromosome compared to the Y chromosome, using a described real-time PCR assay,[2] although 10µg of BSA was added to the reaction to overcome inhibition seen in the Dillema samples. Standard curves were created from triplicate measurements of known DNA samples: XY (male control), XXY (confirmed Klinefelter's control), and XXXY (created by mixing equal quantities of XX female and XY male DNA). Four of Dillema's DNA samples were measured in duplicate in each assay.

Results and Conclusions

Only low quantities of degraded DNA were recovered, as expected, from the material used for sample collection. Autosomal STR genotyping with the Identifiler and PowerPlex 16 commercial STR kits showed a single autosomal profile (Supplementary Tables 1 and 2), consistent with a single non-chimeric donor, in this case presumed to be Foekje Dillema from family accounts. The lack of additional alleles at the autosomal loci also indicated that DNA obtained was from a single donor (presumed to be Dillema), and had not been contaminated with another individual's DNA. The probability that a chimeric individual would display the same allelic profile at all 17 loci examined was estimated at 1.97×10^{-8} , by calculating the probability that two full siblings (as most chimeras are formed from two zygotes) would share all observed alleles. As such, the possibility that Dillema was a chimera can effectively be ruled out, at least for her skin cells.

Although autosomal loci showed approximately equal quantities of each allele (indicating a 1:1 ratio between homologous chromosomes), the *AMELX* and *AMELY* loci from both PCR kits displayed ratios of 2.85-4.5:1. In contrast, XY male control samples had ratios of 0.7-1.3:1 in all amplifications, as expected. Y-chromosomal STR genotyping with the commercial AmpF/STR Yfiler PCR Amplification kit showed a single and complete Y chromosome profile with all Y-STR loci amplified (Supplementary Table 3), and with peak heights between the loci consistent with degraded DNA. The presence of all Y-STRs analysed in allele numbers consistent with a single Y-chromosome indicates that an entire, single and intact Y chromosome was present. X chromosome STR genotyping using the commercial Mentype Argus X-8 PCR Amplification kit provided information about 5 STRs in 4 linkage groups. The remaining 3 loci could not be amplified due to the extremely degraded nature of the DNA samples. Two amplifications showed a second allele at two loci (DXS10074, HPRTB), but these could not be replicated in other amplifications and therefore are

interpreted as artefacts of amplifying low levels of DNA. The consensus profile showed that only a single allele at each locus was amplified (Supplementary Table 4), indicating that only one type of X chromosome was present. Indirect *AMELX/AMELY* quantification from the Mentype Argus X-8 PCR Amplification kit gave an X:Y ratio of 3.63 (2.18 - 5.29), in agreement with *AMELX/AMELY* results obtained from the Powerplex 16 and Identifiler kits.

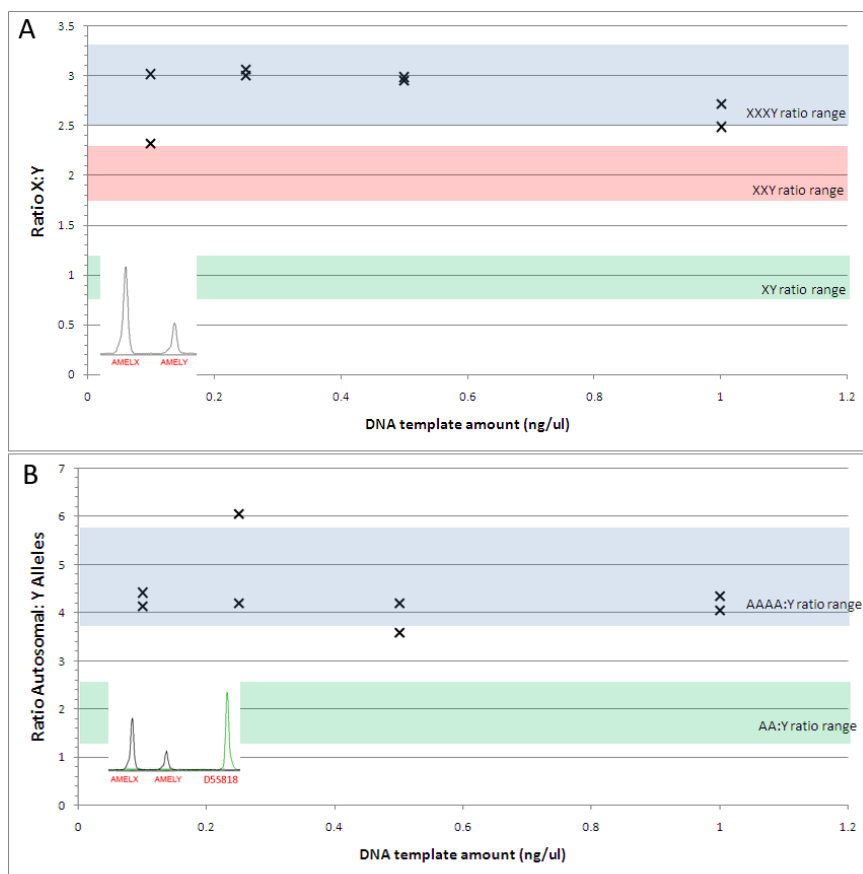
Real-time PCR quantification of the *PCHD11X/PCDH11Y* loci in four of Dillema's samples resulted in a mean ratio of 3:1 for the loci tested, with ratios ranging from 2.32 to 3.66, and six of the eight replicates falling within the XXXY range (Supplementary Figure 1A). Triplicate standard curves gave ratios of 0.8-1.2 for XY controls, 1.76-2.29 for XXY controls, and 2.53-3.39 for XXXY controls (Supplementary Figure 1A). Notably, this ratio, estimated by direct quantitative PCR, was found to be in the range of the more indirect quantification measures for *AMELX/AMELY* obtained via fragment length analysis and peak height inspection (see above). Real-time PCR quantification of autosomal and Y chromosomal loci of Dillema's DNA using the Quantifiler Duo kit gave a 4:1 ratio for the loci tested (Supplementary Figure 1B). Control DNA samples with 1 Y chromosome per autosomal pair gave an average ratio of 1.94 (range 1.37 – 3.01) autosomes per Y chromosome, while controls with 4 autosomes per Y chromosome had ratios of 4.02 (3.23 – 5.98). Dillema's samples ranged from 3.59 to 6.05, with an average of 4.37 (Supplementary Figure 1B), consistent with the presence of one Y chromosome per two diploid sets of autosomes, and an 46,XX/46,XY genotype from a single donor. The presence of a few 45,XO or 47,XXY cells is not excluded, but a significant contribution by such aneuploid cells would be in disagreement with the overall 4:1 ratio.

From the combined genotyping and DNA quantification results, we conclude that Foekje Dillema had a 46,XX/46,XY mosaic genotype, with equal numbers of both genetic cell types at least in her skin, which may have originated from a 47,XXY zygote with one

type of maternal X chromosome. The formation of such a zygote would require fertilization by a normal 23,Y sperm of an aneuploid 24,XX oocyte carrying two sister chromatids from an X chromosome that has not undergone crossing-over in meiotic prophase, followed by nondisjunction of the sister chromatids in the second meiotic division. Alternatively, mitotic nondisjunction might have occurred in a 46,XY embryo, giving rise to a 47,XXY (and the lethal karyotype 45,YO) blastomer. In both scenarios, loss of X and Y by nondisjunction events at mitotic divisions of 47,XXY blastomers must have resulted in clones of 46,XY and 46,XX cells. The above-described events are all known, or at least have been suggested to occur,[4-5] but a series of events leading to a 46,XX/46,XY mosaic individual with one set of autosomes and one type of maternal X chromosome must be extremely rare, as this has been described, to our knowledge, in only two previous case reports.[6-7]

Supplementary References

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Supplementary Data Figure 1.

Sex chromosome and autosome quantification

A. Quantitative real-time PCR targeting the *PCHD11X* and *PCHD11Y* homologous loci.

Triplicate standard curves of XY, XXY, and XXXY DNA were used to define the expected X:Y ratio ranges for each genotype. Six of 8 of Dillema's DNA samples fall within the XXXY range. The inset shows the imbalance between the *AMELX* and *AMELY* peaks in the PowerPlex 16 amplification system, which supports the 3:1 ratio in Dillema's DNA samples.

B. Quantitative real-time PCR targeting an autosomal locus (*hTERT*) and a Y-chromosomal locus (*SRY*). Triplicate standard curves of 46,XY (AA:Y) and 46,XY+46,XX (AAAA:Y) were compared to Dillema's samples, with six of eight samples falling within the 4:1 autosome:Y range. The inset displays the imbalance between the *AMELX* and *AMELY* loci, and an autosomal locus D5S818, further supporting the 4:1 ratio observed.

Table 1. PowerPlex 16 autosomal STR genotypes and consensus profile

sample	D3S1358	TH01	D21S11	D18S51	D5S818	D13S317	D7S820	D16S539	CSF1PO	vWA	D8S1179	TPOX	FGA	Penta_D	Penta_E	AMELX/AMELY
1_08	14,17	7			11	12,14		14,2	12	17,20				14		X
3_08	14,17	6,7	32		11	10,12	12		12	17,20	12	9	23	12,14	10,15	X,Y
5_08	14,17	6,7	28,32	14,17	11	10,12	10,12	12,13	12	17,20	12,13	8,9	21,23	12,14	10	X,Y
10_08	14,17	6,7	28,32	14,17	11	10,12	10,12	12,13	12	17,20	12,13	8,9	21,23	12,14	10,15	X,Y
19_08	14,17	6,7	28,32	14,17	11	10,12	10,12	12,13	12	17,20	12,13	8,9	21,23	12,14	10,15	X,Y
20_08	14,17	6,7	28,32	14,17	11	10,12	10,12	12,13	12	17,20	12,13	8,9	21,23	12,14	10,15	X,Y
consensus	14,17	6,7	28,32	14,17	11	10,12	10,12	12,13	12	17,20	12,13	8,9	21,23	12,14	10,15	X,Y

Table 2. Identifiler autosomal STR genotypes and consensus profile

sample	D3S1358	TH01	D21S11	D18S51	D5S818	D13S317	D7S820	D16S539	CSF1PO	vWA	D8S1179	TPOX	D19S433	D2S1338	FGA	AMELX/AMELY
1_08	14,17	6,7	28,32	14	11	10,12	10,12	12,13	12	17,20	12,13	8,9	14,14.2		21	X,Y
3_08	14,17	7	28		11	10,12		12,13		17,20	12,13	8,9	14,14.2		21,23	X,Y
5_08	14,17	6,7	28,32	14	11	10,12	10,12	12,13	12	17,20	12,13	8,9	14,14.2	18	21,23	X,Y
10_08	14,17	6,7	28,32	14	11	10,12	10,12	12,13	12	17,20	12,13	8,9	14,14.2	18	21	X,Y
19_08	14,17	6,7	28,32	14	11	10,12	10,12	12,13	12	17,20	12,13	8,9	14,14.2	18	21,23	X,Y
20_08	14,17	6,7	28,32	14	11	10,12	10,12	12,13	12	17,20	12,13	8,9	14,14.2	18	21,23	X,Y
consensus	14,17	6,7	28,32	14	11	10,12	10,12	12,13	12	17,20	12,13	8,9	14,14.2	18	21,23	X,Y

Table 3. Yfiler Y-STR genotypes and consensus profile

sample	DYS 456	DYS 389I	DYS 390	DYS 389II	DYS 458	DYS 19	DYS 385	DYS 393	DYS 391	DYS 439	DYS 638	DYS 392	YGATA H4	DYS 437	DYS 438	DYS 448
1_08	16	14			18		14	12	10		23	13	12	15	12	19
3_08	16								10	11						
5_08	16	14	24	31	18	14	11,14	12	10	11	23	13	12	15	12	19
10_08	16	14	24	31	18	14	11,14	12	10	11	23	13	12	15	12	19
19_08	16	14	24	31	18	14	11,14	12	10	11	23	13	12	15	12	19
20_08	16	14	24	31	18	14	11,14	12	10	11	23	13	12	15	12	19
consensus	16	14	24	31	18	14	11,14	12	10	11	23	13	12	15	12	19

Table 4. Mentype Argus X-8 X-STR genotypes and consensus profile

sample	DXS10135	DXS8378	DXS7132	DXS10074	HPRTB	DXS10101	DXS10134	DXS7423	AMELX/AMELY
1_08	20	11		16				15	X,Y
3_08									X,Y
5_08		11		<i>15,16</i>	14			15	X,Y
10_08	20	11		16	<i>13,14</i>			15	X,Y
19_08		11		16	14			15	X, Y
20_08	20	11		16	14			15	X,Y
consensus	20	11		16	14			15	X,Y

Footnote to Tables 1-4:

The above tables display Dillema's STR profiles at a total of 18 autosomal STR loci, 17 Y-chromosomal STR loci, and 8 X-chromosomal STR loci. With reference to Tables 1 and 2, a maximum of two alleles are expected at each autosomal locus. For example, at the locus D3S1358 on chromosome 3, Dillema possessed alleles with 14 and 17 STR repeats (one on the paternal chromosome, one on the maternal). At other loci, the two homologous chromosomes contained STR repeats with the same number of alleles, for example at the locus D5S818 on chromosome 5 both alleles had 11 repeats. For both the Y and X chromosome STRs, only a single allele is expected if a single type of the chromosome is present. In all tables, alleles shown in italics represent drop-in events: alleles could not be replicated in separate amplifications. According to standard interpretation guidelines, these alleles are not included in the consensus profile, as they likely represent random contamination events and/or errors in amplification.