Testicular feminization was the name given to a syndrome found in individuals with female external genitalia and breast development, absent pubic and axillary hair, absent uterus, and intra-abdominal or inguinal testes. It is an X-linked recessive disorder related to an absence of androgen receptors. [The SCISCI database indicates that this paper has been cited in over 245 publications since 1961.]

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"I was working with the late Hans Kottmeier at the Radiumhemmet in Stockholm on an American Cancer Society fellowship when he asked me to see a patient who had been treated for a dysgerminoma of the ovary. The fact that she had no uterus, no axillary or pubic hair, but otherwise appeared to be a normal female, reminded me of a similar case I had encountered while working as assistant to J. V. Meigs at the Massachusetts General Hospital in Boston. Meigs's case was thought to be a true hermaphrodite. I had reviewed the slides on that patient and found tubules and ovarian-like stroma but no follicular elements. Review of the slides in Stockholm showed rudimentary testicular tubules scattered through the dysgerminoma (seminoma).

"By spending long hours in the library, I was able to unearth 80 similar cases in the literature going back to 1817. Some were reported as a special form of pseudohermaphroditism, but others because of their completely feminine appearance were incorrectly assumed to have ovaries, often with germinomas, arachnoidomas, or other tumors. If men as distinguished as Meigs and Kottmeier did not recognize that their patients had this disorder, it seemed worth calling attention to as an entity."

"The selection of a name for the syndrome was a problem. Karyotypes, Barr bodies, hormone receptors, and radioligand assays were unknown at that time. Because patients who were castrated prior to puberty did not get significant breast development, and therefore the testes appeared to be producing estrogens, 'testicular feminization' seemed an acceptable term. The suggestion made subsequently by Lawson Wilkins that the syndrome might be due to androgen insensitivity did not in my mind explain the breast development, until a former Yale resident, O. J. Miller, asked me one day, 'How do you know that you would not have female breasts if they were not suppressed by your testosterone?' My initial negative response was altered when I recalled an untreated case of congenital adrenal hyperplasia who had no breast development in spite of the presence of ovaries because of suppression by her adrenal androgens."

"Further studies carried out with Mahesh, Kase, Vande Wiele, Dorfman, and Lubos showed normal male androgen production and XY karyotypes. Ultimately, lack of androgen receptors was found to be the etiology of the disorder. Griffin and Wilson have recently summarized the work to date on various androgen resistant syndromes. Other reviews have also appeared."

"While this syndrome should therefore be called familial congenital androgen receptor deficiency, we all have our egos. Mine was bolstered when I heard of 17β-male—and even more when my secretary showed me my name in Stedman's Medical Dictionary with the word syndrome attached to it."

"Sexual differentiation is a provocative subject. The original article called attention to a generally unrecognized entity. Although there was an unwillingness on the part of some gynecologists to accept the fact that such completely feminine appearing individuals were XY with tests, the syndrome accounts for seven to ten percent of cases of primary amenorrhea."