Case Report

Plexiform Neurofibroma Presenting as Ambiguous Genitalia

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Abstract

Plexiform neurofibroma rarely affects the external genitalia and is a rare cause of clitoromegaly. The essential aspect of the management is doing a clitoroplasty with the preservation of glandular and neurovascular tissues. We present the case of an 18-year-old female who had ambiguous genitalia due to an enlarged clitoris due to neurofibromatosis that was the size of a penis with indistinct labia majora. She was managed by carrying out a feminising genitoplasty, a combination of clitoroplasty and labiaplasty.

Keywords: Clitoral neurofibromatosis, clitoromegaly, clitoroplasty, feminising genitoplasty

Introduction

Plexiform neurofibroma of the clitoris is a rare condition.[1-3] It grows slowly in adults and older adolescents and may even cease growing altogether unlike in children where the growth rate is faster and may be faster than the overall body growth.[4] The hallmark of surgical treatment of clitoral enlargement is the preservation of clitoral tissue.[5] We report the case of an 18-year-old female who presented with plexiform neurofibroma of the clitoris, absence of the labia majora which is uncommon, and gluteal masses. Her clitoral enlargement was managed with clitoroplasty, and the redundant clitoral tissues were utilised in constructing new labia majora.

Case Report

An 18-year-old female presented to the urology clinic with complaints of ambiguous appearance of the external genitalia from birth. At birth, her mother noticed abnormal external genitalia with an unusual mass at the anterior aspect of the vulva. There was a progressive increase in the size of the mass as she grew up. It was flaccid, did not become erect at any time, and was pendulous by the time she presented to the urology clinic. She had two vulvar openings, an anterior one through which she voids urine and a posterior introitus. In addition, she had multiple macules and papules on the skin that were prominent on the thigh and back. She also noticed painless lumps on her left gluteal area and posterior upper left thigh. There was no history of similar lesions in any member of her family. She is a product of a twin gestation, and the pregnancy was uneventful. Her birth was through spontaneous vertex delivery, and she cried immediately after birth. Her developmental milestones were achieved at the appropriate times. She developed breasts at the age of 12 years, her menarche was at the age of 14 years, and her monthly menstrual cycle was regular. She also had feminine fat and hair distribution. There was no family history of similar condition, infant death, or primary amenorrhea and no history of the use of steroids during her pregnancy.

On examination of the external genitalia, there was a huge mass of tissue occupying the area of the clitoris with what appeared to be the glans of the clitoris at its apex [Figure 1]. The mass measured 13.1 cm × 8.2 cm × 6.3 cm in size. The mass felt doughy on palpation and was not tender. There was a groove of mucous epithelium extending from the vulva toward the summit of the mass giving the impression of a poorly developed urethral plate. There were no palpable gonads in the mass or around the groin. She had another mass on the proximal aspect of the left buttock, close to the natal cleft, measuring 6 cm × 5.5 cm [Figure 2]. Her skin had pigmented macules (café au lait spots) and papular lesions (neurofibromas)

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at the back, gluteus, and upper thigh. A diagnosis of ambiguous genitalia (46 XX DSD) with plexiform neurofibroma of the clitoris was made. She was evaluated and worked up for surgery on the genitals. Her full blood count had values within the normal ranges. She had adequate kidney and liver functions. Abdominopelvic computed tomography scan findings were in keeping with features of normal internal female genitalia with clitoromegaly; there was no testis.

Her surgery was done by a combined plastic surgery team and urology team. We found a pendulous infrapubic fibrofatty mass that was hanging freely toward the introitus. It was occupying the area the clitoris would normally occupy and adjoining contiguous areas. The labia majora were absent on both sides, and the labia minora were poorly defined. The introitus was narrow with a low-lying urethral meatus. A ventral approach was used to avoid injury to the neurovascular bundle. We did a clitoroplasty that preserved the tissues at the apex as the new clitoris and the excess cutaneous flaps were used in reconstructing the labia on either side of the introitus [Figure 3]. This was achieved by making incisions on either side of the vagina, lateral to the poorly developed minora. The proximally based flaps were folded longitudinally, and the edges were sutured to the edges of each incised area. Bleeding was moderate, and there was no need for transfusion. The excised tissue was sent for histopathology, and the report confirmed that it was a plexiform neurofibroma.

Six weeks post clitoroplasty, the patient expressed satisfaction with the outcome of the surgery, rating the current appearance of the vulva as 90% satisfactory. She was yet to resume sexual activity.

Discussion

Plexiform neurofibroma rarely affects the external genitalia[6,7] nor results in clitoromegaly.[2,3] In males, it can present as an enlarged penis.[3,7] Other sites that have been affected by plexiform neurofibromatosis are the cervix, uterus, and ovaries.[3] It has also been reported to affect the vagina[6] and parts of the vulva (mons pubis and labia majora).[3,6] The left gluteal area was affected as well in the index patient.

Most patients with clitoral enlargement present as children. Adolescents and adults presenting with clitoral enlargement usually have low self-esteem and worry about their sexual lives, while some present with psychological distress.[8] We observed a significant improvement in the psychological well-being of our patient after surgery.

The main modality of treatment of clitoromegaly is surgery. The essential aspect of the surgery is clitoroplasty with the preservation of glandular and neurovascular tissues to ensure sensation of the neoclitoris. However, it has been reported that although the neurovascular bundle is preserved at surgery, this may not translate to normal sensitivity of the clitoris afterward.[5] For the index patient, we carried

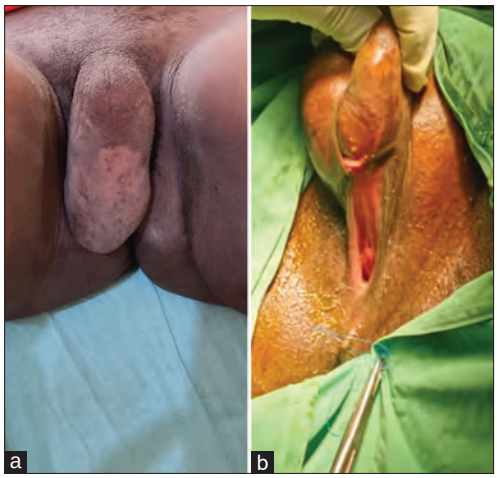

Figure 1: (a) Enlarged clitoris resembling a penis and covering the vaginal opening. (b) The large clitoris raised to expose the vaginal opening

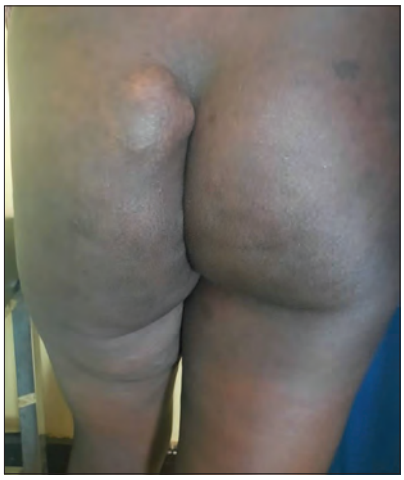

Figure 2: Gluteal plexiform neurofibroma in a patient with neurofibromatosis type 1

Figure 3: (a) External genitalia just after surgery, showing the newly constructed labia majora, urethral catheter and drain. (b) The external genitalia a few days after surgeyy

out a feminising genitoplasty, which is a combination of clitoroplasty and labiaplasty.[5] We chose this procedure because it plays the dual role of addressing the enlarged clitoris and improving the appearance of the vulva by the reconstruction of the labia. The other reason is that the patient identified as a female psychologically; she was raised accordingly and also had features in keeping with internal female genitalia.

Apart from haematoma, which is common with surgeries on plexiform neurofibroma, another complication of surgical excision of this benign tumour is recurrence. However, in older adolescents and adults, the lesion can stop growing or have a decrease in the growth rate.[4] This recognised pattern of progression of neurofibroma suggests that the chances of regrowth of the lesion in our patient who is 18 years are low. It is also recognised that it is extremely rare for clitoral plexiform neurofibroma to undergo malignant transformation.[1,3]

There is still an ongoing debate on whether an early surgery is better than a late surgery or vice versa.[5] The overall prognosis of doing genitoplasty for a patient with plexiform neurofibroma of the clitoris is fair when it is done on adolescents or adults. Decision-making was rather straightforward on this occasion because the index patient presented as an adolescent. We, however, intend to follow up with the patient for some years, and any recurrence would be treated by a revision surgery.

Conclusion

Clitoral enlargement due to plexiform neurofibromatosis is rare. It is best treated by clitoroplasty with efforts made to preserve sensation in the clitoris. In some cases, labiaplasty is also performed to correct distortions in the vulva that may be caused by the large neurofibromas. Surgery is preferably done in older adolescents or adults to decrease the incidence of recurrence.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that her name and initials will not be published, and due efforts will be made to conceal her identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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