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Labioplasty in a Freeman–Sheldon syndrome patient

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Summary
A case of a labioplasty in an 18-year-old Freeman-Sheldon patient with labia minora hypertrophy is described.

Key words: Freeman-Sheldon syndrome; Labia minora hypertrophy; Labioplasty.

Introduction

Freeman–Sheldon syndrome (FSS), also termed distal arthrogryposis type 2A (DA2A), or Whistling-face syndrome, was originally described by E.A. Freeman and J.H. Sheldon in 1938 [1]. FSS is the most severe form of distal arthrogryposis [2]. Patients diagnosed with this condition have characteristic phenotype which comprises of dysmorphic features such as mask-like face, microstomy, philtrum flattening, whistle-like mouth shape, nasal alae hypoplasia, impaired palatial function, and camptodactyly with ulnar deviation and club feet [2]. Anatomical abnormalities can lead to gradual hearing loss, scoliosis, and walking difficulties [2]. In terms of genetic background FSS is caused by mutation in MYH3 gene locus 17p13.1 [3].

The authors describe a case of a labioplasty in an 18-year-old patient with labia minora hypertrophy which is to the present authors' knowledge the first reported case of coincidence of FSS with genital abnormalities.

Case Report

An 18-year-old FSS patient was admitted to the Clinic of Adolescent Gynecology and Sexology of Department of Perinatology and Gynecology of Poznań University of Medical Sciences because of hypertrophy and hyperpigmentation of labia minora. She was diagnosed with FSS in an early childhood and since then she passed several reconstructive surgeries and rehabilitation.

At admission the patient reported discomfort, pain, and physical activity limitation due to labia minora hypertrophy and hyperpigmentation. She reported regular menstrual cycles since menarche at the age of 14, no symptoms of hormonal disturbances, no other chronic diseases or surgeries not related to FSS diagnosis.

At physical examination, development of sex characteristics was adequate to the age according to Tanner's scale: P4, A4, Th4. Gynecological examination revealed labia minora hypertrophy (8 cm length between the base and the wedge), petite vagina, and small opening in the hymen. The uterus and adnexa were normal in transrectal palpation. Transrectal ultrasound showed normal uterus and ovaries. Hormonal tests (folliculotropin, lutropin, prolactin, thyreotropin, total testosterone, and sex hormone binding globulin) were in the normal ranges. Patient was informed about the possibility of labioplasty procedure, accepted the proposal of surgical treatment, and signed the informed consent.

Labial reduction was performed by the wedge resection of labia adjusting the size to the desired one (2 cm length between the base and the wedge). Three weeks and six months after surgery the patient reported full acceptance of her genital anatomy. She did not mention any more the hyperpigmentation of labia and reported successful sexual life with her boyfriend.

Discussion

To the present authors' knowledge, this is a first report of labia minora hypertrophy in a patient with FSS. Thus far, no embryological connection between the labia minora overgrowth and FSS was reported.

Incorrect and unacceptable genital anatomy in adolescents might lead to severe sexual dysfunctions. Labia minora hypertrophy is a source of concerns in adolescent and young women due to functional, esthetic, and emotional reasons [4]. The nuisance of this condition affects especially physically and sexually active girls and women and those who are involved in sports (e.g. horse riding, cycling, jogging, etc.) [4]. The overgrowth can lead to discomfort, abrasions, and trauma in genital area during sitting and walking and hygienic problems, especially during menstruation [5]. Concerning sexual life it may cause dyspareunia and even sexual inhibition [4].

In terms of definition, labia minora hypertrophy can be diagnosed as overgrowth of the inner labia when the distance from the bottom to the edge of the lip is large than 4

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cm [6]. Surgical treatment is suggested for the sake of patients' psychological and physical comfort. In the literature several approaches of plastic surgery of labia minora hypertrophy are described. Before the surgery the exact size of inner labia has to be measured and the result and extent of the procedure should be discussed thoroughly with the patient considering satisfaction after surgery. Patient has to sign consent for the surgery, assenting proposed treatment. Labioplasty is performed by wedge resection of parts of labia adjusting the size to the desired one and by using the circumference reduction and well-planned technique [5-7].

Labioplasty in a FSS patient with labia minora hypertrophy can help to obtain both the full acceptance of genital anatomy and successful sexual life. Multidisciplinary cooperation between specialists is important for treatment all patients with labia minora hypertrophy: it is gynecological and sexual problem. Human sexuality is an important element of the development of the personality [7, 8].

From the ethical point of view, it is fully understandable and permissible to conduct reconstruction surgery after injuries and in illnesses, or to repair deformations stemming from congenital malformations. In some cases it may be even considered necessary or indispensable to undergo plastic surgery. This is particularly true for children with malformations which may in the future stop their personal development. In such situations the burden to recognise the necessity and to take action lies with the parents [7, 8].

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