

# Lipoma of the Clitoris: A Differential Diagnosis within Disorders of Sex Development

## Abstract

**Introduction:** Genital lipomas are a rare type of benign soft tissue tumors that may mimic a disorder of sex development (DSD). Genetic, chromosomal, hormonal and developmental alterations are differential diagnosis that must be taken into account. Consequently, medical staff should have wide knowledge of the possible differential diagnosis regarding DSD since this pathology constitutes a medical and social emergency.

**Results:** A three-year-old female child with a congenital cutaneous superficial lipomatous nevus of the clitoris who arrived to our institution with a misdiagnosis of DSD since birth. Preoperative laboratory analysis, imaging studies and biopsy, lead to proper diagnosis and further surgical removal and clitoridoplasty.

**Conclusion:** Differential diagnosis of DSD includes infrequent pathologies as the one described. Early approach of a multidisciplinary team is essential in the treatment of these patients.

**Keywords:** Female genitalia; Lipoma; Clitoris; Infant; Sexual development disorder

## Case Report

Volume 3 Issue 1 - 2016

**Jaime Francisco Pérez<sup>1\*</sup>, Verónica Tobar<sup>2</sup>, Camila Céspedes<sup>3</sup>, Alejandro Aparicio<sup>2</sup> and Cristina Domínguez<sup>4</sup>**

<sup>1</sup>Department and Pediatric Urology Section, Hospital Universitario San Ignacio and Fundación Santa Fe de Bogotá, Colombia

<sup>2</sup>Urologist, Hospital Universitario San Ignacio, Colombia

<sup>3</sup>Pediatric Endocrinologist, Hospital Universitario San Ignacio, Colombia

<sup>4</sup>General practitioner, Universidad de los Andes, Colombia

**\*Corresponding author:** Jaime Francisco Pérez, Pediatric Urologist, Hospital Universitario San Ignacio, Carrera 7 No. 40-62, Bogotá, Colombia, Tel: (57) 1- 5946161; Email: jaime.perez@javeriana.edu.co

**Received:** September 26, 2015 | **Published:** January 29, 2016

## Introduction

In one of every 4500 to 5000 newborns, appearance of genitals is so abnormal that it is not possible to define the gender of the neonate [1,2]. Findings of disorders of sex development (DSD) include a broad-spectrum group of disease with heterogeneous appearance [3] that should trigger alarms both for parents and medical staff since it constitutes a medical and social emergency [4].

There is a wide variety of differential diagnosis when it comes to pediatric genitoinguinal masses, including hernias, inflammatory process, benign and malignant tumors. Mass characteristics and location should be considered when assessing differential diagnosis. Among the rare cases, it is common to encounter hymenal or paraurethral cysts, hydroceles, perineal lipomas, and genital prolapse at the interlabial area, and among those found at intralabial area, granular cell tumors, fibro epithelial polyps, neurofibromatosis, hemangiomas and hamartomas are most common [5]. Malignant lesions include embryonal rhabdomyosarcoma and endodermal sinus tumors [6]. Consequently, medical staff should have knowledge of all possible differential diagnosis, a suitable diagnostic and therapeutic approach.

The superficial cutaneous lipomatous nevus was first described by Hoffmann and Zurhelle [7], as a dermal mass of adipose tissue that lay in cutaneous folds or in the pelvis and may be solitary lesions; frequently found in newborns until the second decade of life [8], however there is a congenital predominance. Histopathological characteristics are described as ectopic adipose groups over collagen fibers of papillary dermis with no

connection to subcutaneous adipose tissue [9]. We present a female infant with a congenital rapidly growing clitoris mass in whom a diagnosis of cutaneous lipomatous nevus was confirmed after surgical excision.

## Case Report

A three-year-old female with increasing clitoris that doubled its size since birth was assessed at Hospital Universitario San Ignacio, Colombia. She was conceived by a primigravida and non-consanguineous parents with no other significant family or medical history. There were no signs of maternal masculinization or androgenic medication intake during pregnancy, therefore, virilizing tumor or any other exogenous exposure to androgen were both excluded. During antenatal ultrasounds, only the third trimester assessment was able to determine a female gender and no other abnormalities. After an uncomplicated pregnancy, a 37-week female infant was delivered by caesarean section due to cephalopelvic disproportion. The patient was initially diagnosed with DSD soon after birth and a female gender was assigned at a low complexity medical center. On review of symptoms, parents referred normal feeding and meeting all developmental milestones with no delays. No urination or defecation problems, no pain symptomatology or constitutional symptoms.

Physical examination demonstrated generalized increase of body and glans of the clitoris with 4.5 cm length and 2 cm in width on gross examination (Figure 1). It was not possible to identify preputium clitoridis or urogenital sinus; adjacent were normal labia majora and minora, introitus and urethra meatus with no signs of virilization (Figure 2). Physical examination was otherwise unremarkable.



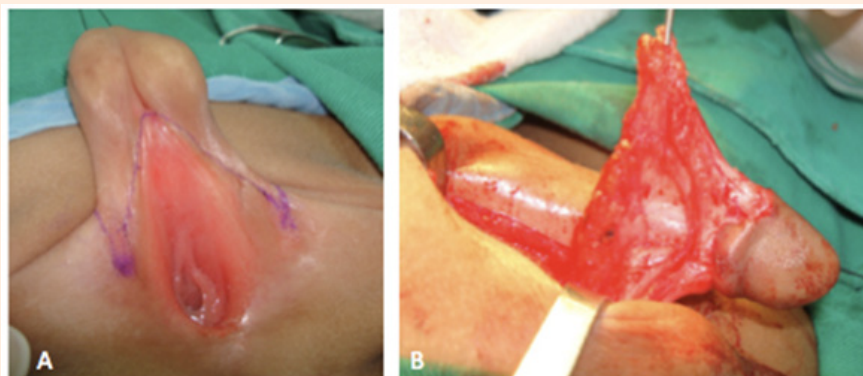
**Figure 1:** Generalized growth of the clitoris body and gland of 4.5 x 2 cm of diameter.



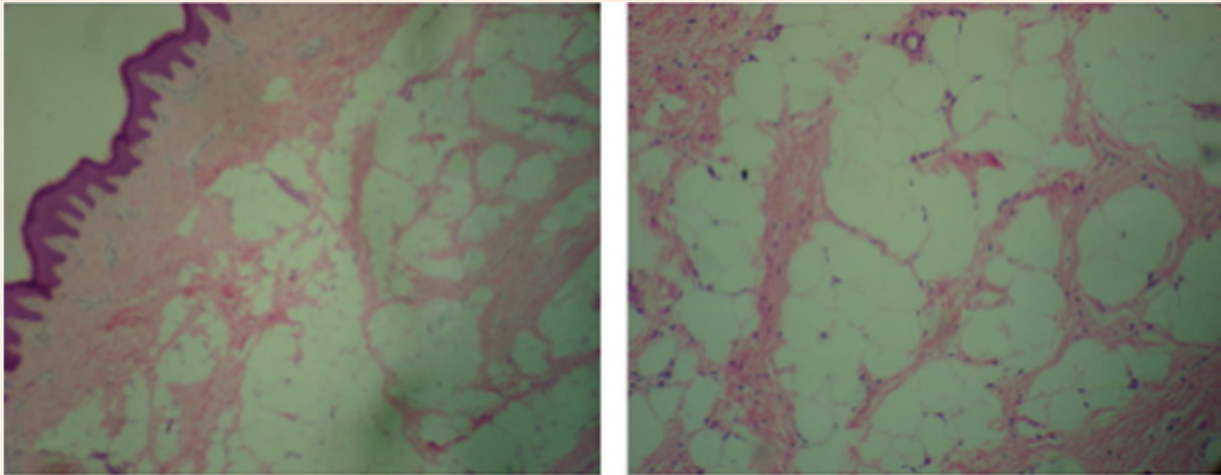
**Figure 2:** Normal labia majora and minora, introitus and urethra meatus.

Further studies to discard feminine hermaphroditism were evaluated. Laboratory tests documented 46, XX karyotype on 25 studied metaphases; normal 17-hydroxyprogesterone, androgen levels, androgenic precursors and hypothalamus-pituitary-gonadal axis function. Pelvic ultrasound confirmed female organs and the color Doppler of external genitalia identified nonspecific prominence of the clitoris with no mass or vascularization identified. The patient had a normal cystoscopy, vaginoscopy and biopsy findings were consistent with a cutaneous and subcutaneous lesion, characterized by overgrowth of mature adipose tissue, compatible with diffuse lipomatosis. With these results plexiform neurofibroma and hamartoma were excluded.

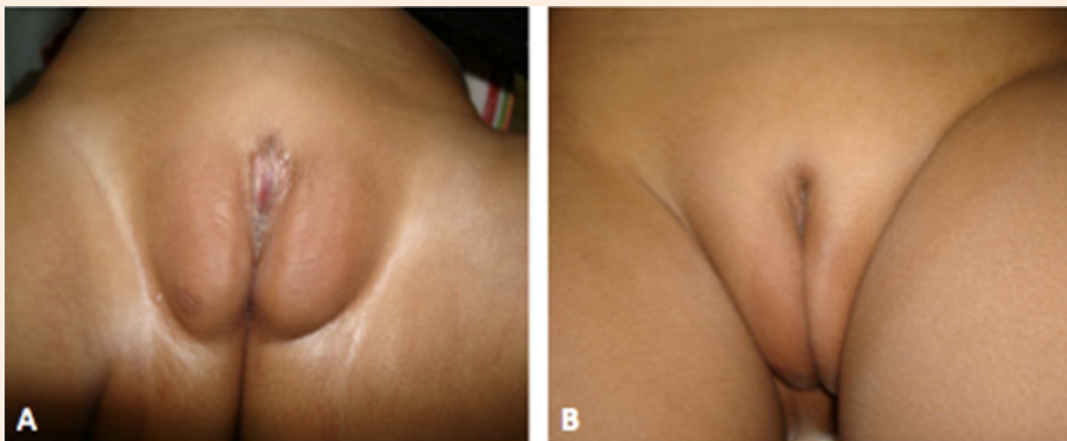
The patient underwent mass resection and clitoridoplasty. Surgical findings reported a dependent lesion in prepubic area, at the level of the suspensory ligament and extending to the tip of the clitoris (Figure 3a). Tunica albuginea and neurovascular bundles were identified and preserved (Figure 3b). Final pathological analysis reported superficial cutaneous lipomatous nevus (Figure 4). The patient was followed in the clinic postoperatively for 10 months with adequate medical progress, without signs or symptoms of recurrence and currently with a normal female genitalia appearance (Figure 5a & 5b).



**Figure 3:** a) Dependent lesion of prepubic area at the level of the suspensory ligament and extension to the tip of the clitoris. b) Tunica albuginea and neurovascular bundles.



**Figure 4:** Haematoxylin and eosin stain, transverse cut of mass showing a lipoma with mature white adipocytes and numerous capillary blood vessels. No immature, atypical adipocytes or mitotic signs.



**Figure 5:** a). Two weeks postoperative with adequate medical progress. b). 10 months later with no recurrence and normal female physical appearance.

## Discussion

Genital masses in pediatric population are infrequent and in occasions can mimic a hypertrophy genital tubercle leading to erroneous diagnosis of DSD. They have a wide variety of characteristics and therefore differential diagnosis include those of benign and malignant lesions as condylomas, hymeneal or paraurethral cysts, hydroceles, capillary or cavernous hemangiomas, lipomas, lymphangiomas, neuro fibromas, embryonal rhabdomyosarcoma or endodermal sinus tumors. Supernumerary glands derived from the embryonary mammary line are also rare causes of vulvar masses [10].

Lipomas are the most common soft tissue tumor of adulthood but they are rare during the first two decades of life; less than 5% are diagnosed in this population [11]. Most congenital perineal lipomas occur in male infants and present as accessory scrotum

[12]. Also, they have been reported as hamartomas instead of true neoplasms [13]. Our patient had a true congenital lipoma that had a progressive growth during the first two years of life. Lipomas are well-encapsulated adipose masses that may be found at multiple parts of the body as single, multiple, superficial or deep lesions. There is no histological difference between normal adipose cells and those found in the lipoma, however, lipomas contain larger amount of protein and lipase [14]. When identified in the vulvar area, lipomas present as prominences of the labia mayora and are most frequent in older women [15]. Adenolipomas are a rare variant that may compromise external genitalia but are also found in women from 25 to 75 years of age. The pathognomonic sign of this lesion is the presence of eccrine glands within them [8]. The principal differential diagnosis of the external genitalia lipoma is the plexiform neurofibroma which may produce clitoromegalia in patients with Von Recklinghausen disease [8]. Etiology of genital

masses can be differentiated with gonadotropins, androgens, ultrasound imaging and karyotype studies that can assist in the differentiation of DSD and ambiguous genitalia.

The term lipomatosis has been used to describe cutaneous, subcutaneous, multiple or solitary lesions, or to name the diffuse neoplastic lesion characterized by adipose tissue proliferation [16]. It is three times more common in men and can be associated as well as lipomas, with hypercholesterolemia. Lipomatosis in childhood can be related to development alterations or mesenchymal malformations instead of neoplastic lesions; some lipomatosis subtypes include congenital infiltrative lipomatosis, superficial cutaneous lipomatous nevus, proteus syndrome, angiomatosis-lipomatosis and macro-encephalic syndrome [17].

Superficial cutaneous lipomatous nevus is a rare condition characterized by ectopic adipose tissue accumulation that is located almost always on the lower trunk [18]. In some cases, this type of lesions can appear as masculine genitalia and required further evaluation for a precise diagnosis and treatment. Our patient lived for three years with a misdiagnosis of genital ambiguity until she received a multidisciplinary approach that included urology and pediatric endocrinology, which lead to the correct diagnosis of a lipoma subtype and received adequate surgical correction.

## Conclusion

Differential diagnosis of DSD includes infrequent pathologies as the one mentioned in this case. Early multidisciplinary medical approach is indicated for these patients and should be managed at high complexity medical centers. Laboratory analysis of gonadotropins, androgens, karyotype analysis and radiological imaging studies such as ultrasonography are useful for making an accurate diagnosis. Surgical excision and in this case clitoridoplasty are the treatments of choice.

## References

- Hamerton JL, Canning N, Ray M, Smith S (1975) A cytogenetic survey of 14,069 newborn infants. I. Incidence of chromosome abnormalities. *Clin Genet* 8(4): 223-243.
- Allen L (2009) Disorders of sexual development. *Obstet Gynecol Clin North Am* 36(1): 25-45.
- Mahalik SK, Mahajan JK, Sodhi KS, Garge S, Vaiphei K, et al. (2013) Rare association in a female DSD case of phallus, accessory phallic urethra, perineal lipoma and anterior ectopic anus. *J Pediatr Urol* 9(1): e39-42.
- Brown J, Warne G (2005) Practical management of the intersex infant. *J Pediatr Endocrinol Metab* 18(1): 3-23.
- Kirkham YA, Yarbrough CM, Pippi Salle JL, Allen LM (2013) A rare case of inguinolabial lipoblastoma in a 13-month-old female. *J Pediatr Urol* 9(1): e64-67.
- Ambros R, Kurman R (1992) Tumors of the vulva. In: *Pediatric and Adolescent Gynecology*. Raven Press, New York, USA, pp. 353-363.
- Hoffmann E, Zurhelle E (1921) Übereinneaevus lipomatous cutaneous superficial is der linken Glutaalgegend. *Arch Derm Syph* 130: 327-333.
- Pantanowitz L, Henneberry JM, Otis CN, Zakhary M (2008) Adenolipoma of the external female genitalia. *Int J Gynecol Pathol* 27(2): 297-300.
- Hattori R, Kubo T, Yano K, Tanemura A, Yamaguchi Y, et al. (2003) Nevus lipomatous cutaneous superficialis of the clitoris. *Dermatol Surg* 29(10): 1071-1072.
- van der Putte SC (1994) Mammary-like glands of the vulva and their disorders. *Int J Gynecol Pathol* 13(2): 150-160.
- Coffin CM, Dehner LP (1990) Soft tissue tumors in first year of life: a report of 190 cases. *Pediatr Pathol* 10(4): 509-526.
- Numajiri T, Nishino K, Sowa Y, Konishi K (2011) congenital vulvar lipoma within an accessory labioscrotal fold. *Pediatr Dermatol* 28(4): 424-428.
- Jacobi A (1884) Congenital Lipoma. *Arch Pediatr* 1(11): 65-83.
- Coffin C, Dehner L, O'Shea P (1997) Pediatric soft tissue tumors. In: Williams and Wilkins (Eds.), *A clinical, pathological, and therapeutic approach*, USA.
- Van Glabeke E, Audry G, Hervet F, Josset P, Gruner M (1999) Lipoma of the preputium clitoridis in neonate: an exceptional abnormality different from ambiguous genitalia. *Pediatr Surg Int* 15(2): 147-148.
- Weiss S, Goldblum J (2008) *Enzinger and Weiss's Soft Tissue Tumors*. (5<sup>th</sup> edn), Mosby, USA, pp. 581.
- Gilbert-Barness E (2007) *Potter's Pathology of the Fetus, Infant and Child*. (2<sup>nd</sup> edn), Mosby, USA.
- Stenger D, Bahmer FA (1986) Superficial cutaneous lipomatous nevus (Hoffmann-Zurhelle)]. *Z Hautkr* 61(1-2): 50-56.