Summary

A 14 year old girl was seen in the clinic with a huge vulvar wart, and warts on both tonsils. She also had poikiloderma. She had bony abnormalities, which included lobster claw abnormality of the right foot, a right cervical rib, and the right clavicle was lower than the left. There were fine parallel vertical radio-opaque lines in the distal femoral tibial metaphyses bilaterally (osteoapathy striata).

A diagnosis of Focal dermal hypoplasia was made. The huge vulvar wart and the warts on the tonsils were excised. She recovered promptly and she was discharged home.

Keywords: Focal dermal hypoplasia, Nigerian, Case report

Résumé

Une fille âgée de 14 ans était vue dans la clinique atteinte d’un défaut énorme dans la vulve, et des déformations sur les deux amygdales. Elle est également atteinte de la poikiloderme. Elle a mal dans l’os y compris une anomalité dans le loboter claw de la jambe droite, une côte cervicale du droit et la clavicule du droit plus basse que celle de gauche. Il y a des lignes radio-opaque fines parallèles verticalement dans les métaphyses distale femorale (ostéopathie striata).

On a fait un diagnostic de la hypoplasie dermique focale. Le défaut très énorme de la vulve et les défauts sur les amygdales ont été opérés. Elle s’est rétablie rapidement et sortie de l’hôpital.

Introduction

Focal dermal hypoplasia is an uncommon disorder affecting organs derived from the ectoderm and mesoderm. Its cutaneous features were first described in 1921 and the non-dermological components of the disorder were highlighted by Golitz in 1962. One of the unique features of the disorder is its variability in organ involvement. The skin, eyes, and skeletal tissue are usually involved. One of the components of the syndrome, papillomas may be present at birth or appear at different sites later on in life. The patients may present with life threatening situations e.g. papillomas may develop in the larynx requiring tracheotomy. The papillomas may also mimic infections such as genital warts which may be wrongly attributed to child abuse in children. We present a case of FDH in a 14 years old Nigerian female who presented with papillomas on the tonsils and perianal area. She also had a capillary haemangioma in the right gluteal region. As far as we know haemangioma has not been previously documented in this syndrome and it is the first case reported in a Nigerian.

Case report

A 14 year old Nigerian girl was seen in the Gynaecological department of the University College Hospital Ibadan with a five month history of a vulvar mass which was progressively increasing in size. The mass bled especially when it was traumatized and exuded foul smelling fluid. She denied any history of sexual intercourse in the past. Her mother had noticed that she snored during sleep and her voice had become somewhat muffled in the last two years.

There were no associated systemic symptoms and the patient felt well otherwise. Her past medical history revealed that she had presented to the same hospital at the age of 4 years with a complaint of a right gluteal mass, which had been present since birth but had increased in size at that time. The mass had been excised and the histology report from the notes and review of the slides at this visit showed the mass to be a capillary haemangioma (Fig. 1). There had been no recurrence of the lesion. She was also noticed to have shortening of the right limb at that visit.

Table 1 Skeletal abnormalities in FDH

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<tr>
<th>Abnormality</th>
<th>Description</th>
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<tr>
<td>Short stature</td>
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<td>Asymmetry of skull, trunk and</td>
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<td>extremities</td>
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<td>Hypoplastic or absent digits</td>
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<td>Syndactyly</td>
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<td>Osteoporosis1,2,3,4</td>
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<td>Fibrous dysplasia1,2,3</td>
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<td>Cyst in metatarsals, tibia,</td>
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<td>tibia, maxilla, ischium</td>
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<td>Widening of pubic symphysis1,2,3</td>
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<td>Abnormalities of ribs and clavicles</td>
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<td>Lobster claw hand and foot</td>
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<td>Vertebral abnormalities e.g.</td>
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<tr>
<td>scoliosis</td>
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<td>Osteopathea striata1,2,3,4,5</td>
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<td>Osteochondroma1,2,3</td>
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and part of the face were affected. The pigmented abnormalities were interspersed with striate atrophic areas and the right side of the body was more affected. (Fig. 2) Erythema was prominent in the hypopigmented area of the lower limbs. She had scarring alopecia in areas affected by the atrophic lesions. The third fingernail was absent and this had been so from birth.

The breasts were normally developed for her size but she had sparse axillary and pubic hair. She had scoliosis and shortening of the right tibia and fibula by about 6cm. She also had a lobster claw deformity of the right foot.

Eye examination revealed posterior subcapsular cataract.

Oral examination revealed small conical shaped teeth with malocclusion.

She had markedly enlarged tonsils with wart-like growths that appeared to meet in the midline “kissing tonsils” (Fig. 3). The larynx appeared normal.

Examination of her genitals revealed a huge right vulvar growth extending to the perianal area with a lobulated surface. It had a few ulcers on the surface and exuded blood-stained foamy serous fluid. Her hymen was intact. The rectal examination was normal.

Examination of the cardiovascular, respiratory, and central nervous systems appeared normal. Results of investigations are outlined below.

Her haematological work up and electrolytes and urea were within normal limits. X-ray findings were rounded lucencies in the
pialcal bones in keeping with congenital pialcal foramina. Both frontal sinuses were dysplastic, while the ethmoids were closely
with the right nasal turbinate engorged, suggesting chronic sinusitis. There was calciﬁation of the left second upper molar and rarefaction of the
maxillary bone anteriorly. There was a right cervical rib and a prominent transverse process on the left side, with the left clavicle being higher than the right (Fig. 4). X-ray of the thorax and knees showed linear lucencies in the muscles of the thighs especially on the
posterior aspect. There were ﬁne parallel vertical radio-opaque lines in the distal femoral and proximal tibial metaphyses bilaterally,
better demonstrated on the right side, osteopaphia striata (Fig. 5). There was also loss of parallelism of the articular surfaces of the
right knee due to uneven modeling and expansion of the medial notch. Pelvic ultrasound revealed a hypoplastic uterus.

She then had diathermy excision of the vulvar growth and bilateral tonsillectomy. Histology of the tonsils and vulvar mass showed acanthosis, papillomatosis and koilocytosis of the epitheliunm with focal parakeratosis. Some of the tonsillar crypts were covered by debris and foamy histiocytes. These lesions were thought to be hamartomas.

Histology of the atrophic skin lesion showed loss of rete pegs with bundles of mature adipocytes within the reticular and papillary dermis associated with atrophy of dermal appendages (Fig. 6). These features were consistent with atrophy that could be associated with Goltz Syndrome. The patient subsequently had an eventful recovery from the surgery and was discharged home.

Discussion
The term ‘focal dermal hypoplasia’ does not describe the protean manifestations of this disorder that involves various organs in the body but it has come to stay. FDH is a developmental disorder of organs of mesodermal and ectodermal origin. There are about 200 cases in the literature. There is strong evidence that it is inherited as an X linked dominant condition. It has a female preponderance and there is an increased frequency of miscarriage of male fetuses in affected females. However a few cases have been documented in males and these have been attributed to half chromatid mutations or autosomal dominant inheritance affecting the germ cells. Spontaneous cases resulting from mutation do occur as can be assumed to be the case in our patient with no family history of the disorder nor mosaicin of it.

The variability in the expression of FDH within families and involvement of the skin and bone can be explained by the Lyon hypothesis in which random inactivation of the mutant gene results in mosaicism, and the percentage of the active mutant X chromosome present would determine the clinical picture in the individual. The atrophic linear streaks with pigmentary abnormalities following Blashko’s lines on the skin, and the linear streaks on the bone indicate some form of X chromosome mosaicism resulting in clonal proliferation of two functionally different populations of cells during embryogenesis.

Some authors have conﬁrmed the ﬁndings of two different populations of dermal ﬁbroblasts in the affected individual with ﬁbroblasts in the affected skin being abnormal while that in the unaffected skin was normal. FDH manifests itself in different organs of ectodermal and mesodermal origin but its effect is mainly on the connective tissue of bone and skin. Patients are of small stature, they have a triangular face and large ears for the size of their face. Cutaneous manifestations of FDH in the same individual may be varied. It may be bilateral but asymmetric or solely unilateral. There are reports of patients with FDH without skin involvement but skin involvement is usually regarded as essential for diagnosis.

When hair and nail disorders are excluded, the major cutaneous features could be grouped into five:

1. Congenital aplasia of the skin
2. Multiform patterns of atrophic like lesions
3. Striata lesions.
4. verrucous papilloma of skin and mucous membrane (angi6fibroma).
5. Lipomatous lesions

Telangiectasia and hyperpigmentation of various design and amounts may accompany or overlap the multiform atrophic lesions and striated lipomatous lesions.

Congenital aplasia of the skin
This is not very common and the skin is completely absent in the affected site at birth. In some patients there may be some bullous eruptions or inﬂammatory process on the skin at birth which usually heals up living a scar as was reported in our patient.

Multiform pattern of atrophic like depression
This has been described with different names such as atrophy, depressions, scas, congenital linear macular atrophederm or focal dermal hypoplasia. Histological features of these lesions depend on the age of the lesion. New lesions may show an increase in dermal blood vessels but the older lesions show a thin or absent dermis, i.e. dermal hypoplasia with mild dermal fat deposits.

Striata lesions
These are linear stripes or streaks which are congenital. They may be inﬂammatory at the onset and may be conﬁned with incontinencia pigmenti. The histology of Striata lesions usually reveals increased proliferation of blood vessels, increase in ground substances, and inﬁltration of lymphocytes and histiocytes.

Papillomas
The papillomatous lesions known as angi6fibroma or ﬁbrovascular papillomas are an important component of the disorder that may be present at birth or develop as the child gets older. They have been reported in various locations in the body but the commonest sites are the perineum, vulva, and perianal regions. The vulvar lesions are usually misdiagnosed as genital warts (condyloma acuminate) as was the case in our patient. The warts on her tonsils had suggested the possibility of oral sex which in the younger patients may incite sexual abuse and lead to false accusations. Papillomas have been reported in the larynx, eye lid margins, mouth, pharynx, tonsils, palate, gums, tongue and lips. The fingers and pinna may also have papilloma on them. These lesions may also produce life threatening situations e.g. papillomas in the larynx requiring tracheotomy, or these occurring the oesophagus causing stiucture. Malignant transformation of papillomas has also been reported. Histology of the papillomas show a ﬁbrovascular stalk covered with a layer of acanthotic stratiﬁed squamous epithelium resembling epidermis with extensive papillary folds. Hyperkeratosis and parakeratosis may be present. The stalk is composed of loose connective tissue with dilated vessels and an admixture of inﬂammatory cells.

Lipomatosis
These manifest as skin coloured nodules or nodules slightly darker than the normal skin. Lipomatosis does not occur in all patients and was not present in our patient. The histology of the lesions show lobulated fat approaching the epidermis but separated by a few strands of connective tissue. This results from hypoplasia of the dermis which varies in the different areas. The hair may be sparse especially if the scalp is involved and apocrine gland abnormalities have been reported. Hydrocystomas have also been reported around the eyes. Pial abnormalities include dystrophic nails or total absence of the
Focal dermal hypoplasia: a case report and review of literature - Ogundibi A. O. et al

Skeletal abnormalities in the patients with FDH have been highlighted in Table 1. Of note is the linear streaks in the metaphysis of the long bones osteopathia straita, which has established itself as one of the hallmarks of FDH.16,27 which was also seen in our patient. It is not present in all cases. The lobster claw deformity affecting the hands or foot are also striking abnormalities of the syndrome. A number of abnormalities of soft tissue have also been reported in a few of these patients. They include urinary tract abnormalities (e.g. holf shoe kidney, dilated ureters with inflammation and cystitis). Others include a diaphragmatic hernia and reflux oesophagitis. Some of the patients do mature and have reproductive capabilities but our patient had a hypoplastic uterus.

Excellent reviews on eye signs in FDH have been published28-30 and they include colobomias, (especially of the iris), microphthalmia, anophthalmia, hypertelorism, optic atrophy, keratoconus, cortical and subcapsular cataract. Strabismus, nystagmus and disorders of lacrimal gland apparatus have also been documented.

Many patients with FDH have some level of mental deficiency and there are reports of meningomyelocele, hydrocephalus and Arnold Chiari malformation in a patient.31 Seizures have not been reported in association with this disorder. Our patient had an associated capillary haemangioma of the right gluteal region which is in keeping with mesodermal abnormalities. As far as we know this has not been previously reported in association with this syndrome.

References