Localizador: 18007

Medicina Cutánea
Ibero-Latino-Americana

Basaloid follicular hamartoma with abnormalities in the nervous, gastrointestinal and musculoskeletal system

Hamartoma folicular basaloide con anormalidades en el sistema nervioso, gastrointestinal y músculo esquelético

Alexandra Mancheno-Valencia,* Laura Gabriela Flores-Peña,‡ Sonia Toussaint-Caire,§ Marisol Díaz-Lozano,§ Adriana Rosas-Manzano,‖ Verónica Fonte-Ávalos,* María Elisa Vega-Memije§

Keywords:
Basaloid follicular hamartoma, Happle Tinschert syndrome, dermatopathology.

Palabras clave:
Hamartoma folicular basaloide, síndrome de Happle Tinschert, dermatopatología.

Abstract

Cutaneous syndromes associated to malformations or pathologies of other organs are relatively common, but diverse with regard to their manifestations. In cases with uncommon features the diagnostic process may be difficult, and the histopathological analysis could be the cardinal step in order to determinate the correct diagnosis. We report the case of a man with a disseminated dermatosis affecting the right side of the body, characterized by brown papules linearly arranged in association to extracutaneous defects (leiomyomatosis peritonealis disseminata, bilateral brachydactylia, shortening of the right pelvic member, genu varum, hypoacusia). A histopathological study was performed showing findings compatible with the diagnosis of basaloid follicular hamartoma. Posteriorly he was diagnosed of symptomatic epilepsy. Basaloid follicular hamartoma is a rare follicular malformation that has been described as an isolated entity or in association with systemic alterations. Previous reports have described cases with similar characteristics to the one we are subjecting, and there is a proposition to group these cases as part of a spectrum of a distinct phenotype.

Resumen

Los síndromes cutáneos asociados con malformaciones o patologías de otros órganos son relativamente comunes, pero diversos en cuanto a sus manifestaciones. En casos con hallazgos poco comunes, el proceso diagnóstico puede ser difícil, y el análisis histopatológico podría ser el dato cardinal que permita llegar al diagnóstico correcto. Se reporta el caso de un hombre con una dermatosis diseminada que afectaba el hemicuerpo derecho, caracterizado por neoformaciones de aspecto papular con una disposición lineal, en asociación a defectos extracutáneos (leiomiomatosis peritoneal diseminada, braquidactilia bilateral, acortamiento del miembro pélvico derecho, genu varum, hipoacusia). Se realizó un estudio histopatológico el cual demostró hallazgos compatibles con el diagnóstico de hamartoma folicular basaloide. Posteriormente el paciente fue diagnosticado de epilepsia sintomática. El hamartoma folicular basaloide es una malformación folicular infrecuente, que ha sido descrita como una entidad solitaria o asociada con alteraciones en otros sistemas. Reportes previos han descrito pacientes con características similares al que nos referimos en este artículo, y existe una propuesta de encasillar estos casos como parte de un espectro de un fenotipo específico.

* Dermatologic Surgery.
‡ Genetic.
§ Dermatopathology.
‖ Dermatology Division.

«Dr. Manuel Gea González» General Hospital, Mexico City.

Conflicto de intereses: Ninguno.

Received: 5/February/2018.
Accepted: 8/April/2019.

INTRODUCTION

Epidermal nevus syndromes are a group of alterations that present malformations limited to the skin with a predominantly epidermal component. They can be observed whether isolated, or in association with systemic alterations.[1] They are classified based on their main anatomic component which can be sebaceous, apocrine, eccrine, follicular and keratinocytic.[2] In many situations the clinical characteristics may not be clear enough to determine a diagnosis, and the hystopathological analysis constitutes an extremely important tool to assess conclusive identification of the disease.

CASE PRESENTATION

A 38-year-old man presented with a widespread dermatosis in the right side of the body (post auricular, trunk, upper and lower limb), following a linear disposition, characterized by multiple brown papules

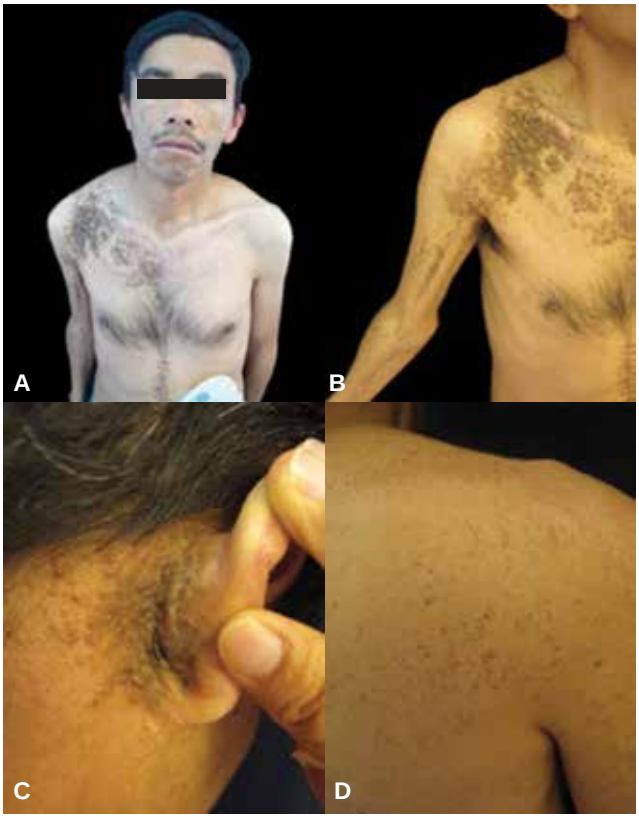

Figure 1. A) Clinical aspect of the basaloid folicular harmatoma. B) The skin biopsy was taken of a right arm lesion. C and D) Clinical details of the lesions.

of various sizes, with well-defined borders, verrucous; some grouped *(Figure 1)*. Hands and feet showed areas of linear atrophoderma with hyperpigmentation *(Figure 2)*. It was noted thick, dry hair with lower implantation in the posterior cervical region, peculiar facies with short palpebral fissures, irregular scattered eye brows, bilateral epicanthus, and trichiasis; high palate, short neck. The thorax presented asymmetrical, with prominence of the left side, and also ipsilateral hypertrichosis. He had a left iliac fossa colostomy because of a recent surgery that revealed an ulcerated vermiform appendix, and a tumor located at the descending colon, with benign characteristics and approximately 2,000 g, diagnosed as leiomyomatosis peritonealis disseminata. He presented small hands with bilateral brachydactylia, shortening of the right pelvic member, *genu varum*, and a surgical scar in the first toe of both feet *(Figure 2)*. He referred also slightly diminished hearing with the right ear and learning difficulties.

Genetic department determined a 46XY karyotype and a renal ultrasound reported changes secondary to an inflammatory renal process. A biopsy of the skin from a papule in the right arm found a lesion emerging from the epidermis coming up to the middle reticular dermis with a proliferation of basaloid and squamous cells, organized in thin strings that were anastomosed, giving a reticulated appearance, surrounded by soft stroma and squamous eddies *(Figure 3)*. It was not found necrosis or atypia. Such findings were compatible with the diagnosis of basaloid follicular hamartoma (BFH).

Eight months after his first dermatological consultation the patient developed tonic convulsive cronic generalized crisis. A cranial axial computerized tomography showed left cerebral hemiatrophy, and symptomatic epilepsy was diagnosed; treatment started with 100 mg oral diphenylhydantoin every 8 hours, a dose that has been retained until now.

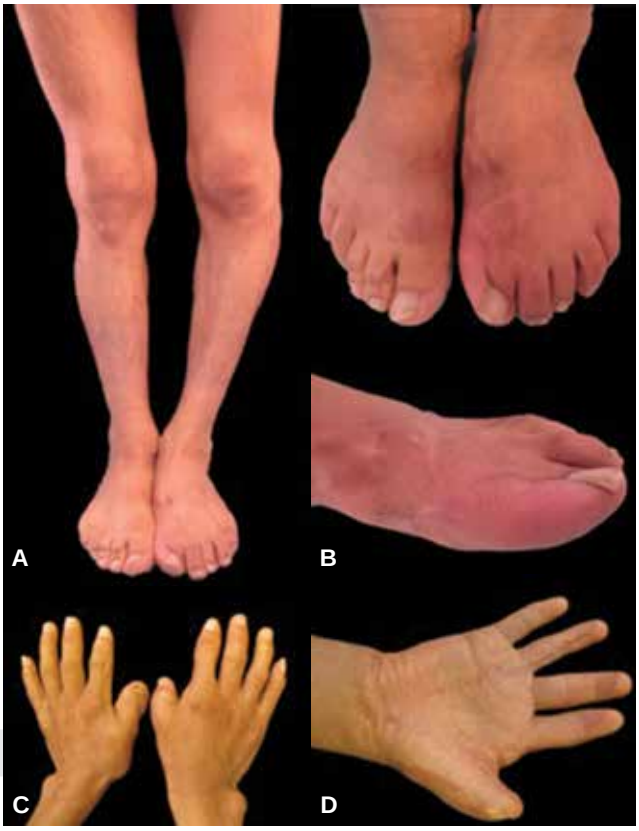

Figure 2. A) Osseous alterations: genu varum, shortening of the right pelvic member. B) Stigmas from previous surgeries. C) Brachydactyly of both thumbs. D) Linear atrophoderma in hands and feet.

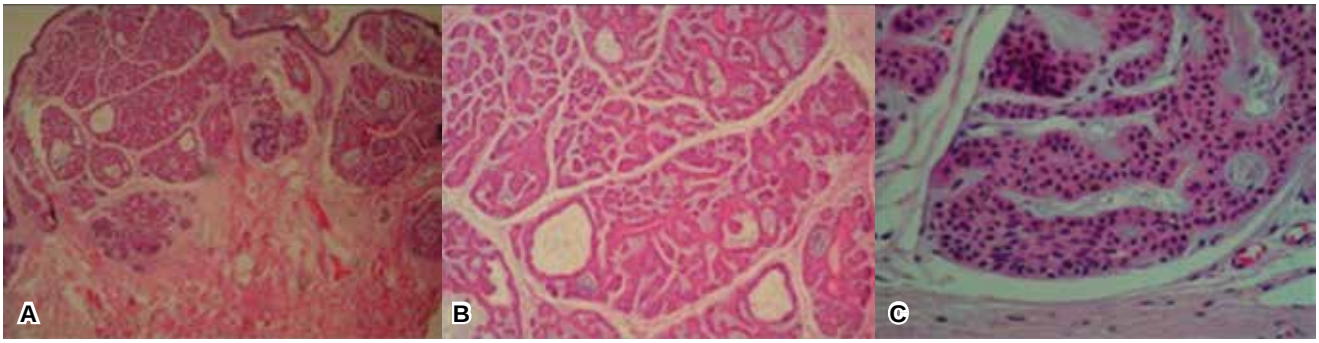

Figure 3. Histological study of a verrucous lesion. Basaloid cells displayed in thin and reticulated strings, surrounded by a fibrous stroma and small keratinous cysts. H&E A) 20x, B) 40x, C) 60x.

DISCUSSION

The patient exhibited a great variety of alterations in different organs. The background of acute abdomen allowed the finding of leiomyomatosis peritonealis disseminata, which in association to the dermatological, skeletal and neurological manifestations led us to propose an epidermal nevus syndrome (ENS). The ENS has been used to describe the association of an epidermal nevus with systemic findings that may affect multiple organs, including neurological, optical, skeletal, and less frequently, cardiac and kidney abnormalities.[3] Certain clinical similarities can complicate the process to get the precise diagnosis; however, particular details of each case allow its correct differentiation.

In this case, the finding of a BFH in the right side of the body was of utmost importance. Histopathology showed its main characteristics (folliculocentric proliferation of uniform basaloid cells that are arranged in cords and clusters and separated from each other by an organized fibrous stroma).[4]

BFH is a rare follicular malformation that exhibits a characteristic histological pattern.[5] Its clinical appearance is heterogeneous, and can be congenital or acquired,[6] showing in some cases a relation to other clinical entities.

It can cause focalized or widespread disease[4] and five clinical forms have been determined: 1) An acquired type, generalized and associated to myasthenia gravis and diffuse alopecia.[7] 2) A generalized type, without related diseases.[8] 3) A congenital type, generalized, associated to cystic fibrosis.[9] 4) A localized, linear and unilateral type.[10,11] 5) A solitary plaque or nodular type.[5] In some cases autosomal dominant inheritance has been described.

It affects both children and adults, and it can be found at any part of the body. The dermatosis can be characterized by neoformations of papular aspect, and comedones or plaques.[12]

Association of BFH to other syndromes has been reported, and in the literature it is possible to find other case reports of patients with similar characteristics to the one we are subjecting. It is notable the analysis made by Rudolf Happle and Sigrid Tinschert[12] who presented a case of a 39-year-old male patient with multiple BFH in a systematized pattern following Blaschko's lines along with osseous, dental, and cerebral anomalies. They also summarized 8 case reports found in the literature that seem to share similar features corresponding to a spectrum of a distinct phenotype.[12]

In 2009, Itin PH[13] reported a case of a 7-year-old patient HBF, enamel defects, webbing of the first and second right toes and medulloblastoma; in his article he proposes to name this condition as Happle Tinschert syndrome.

Our patient could be considered to match this syndrome; however the other defects (leiomyomatosis peritonealis disseminata, hypoacusia, symptomatic epilepsy) could lead as to contemplate other infrequent diagnostic probabilities.

The aims of this case report are to add a new case to the ones catalogued as Happle Tinschert syndrome, or well expose it in order to consider different diagnostic possibilities based on the experience of other authors.

CONCLUSIONS

BFH is a follicular malformation that could share clinical similarities to an ENS. It is of utmost importance to corroborate the histology of the lesions, in order to recognize

the true nature of the disease. These syndromes could be difficult to categorize, and requires the participation of a multidisciplinary team for its management.

Correspondence:
Alexandra Mancheno Valencia
Dermatology Division,
«Dr. Manuel Gea González»
General Hospital, Mexico City.
Calzada de Tlalpan No. 4800,
Colonia Sección XVI, CP 14030, CDMX.
Telephone: 4000 3057
E-mail: aleja_mv@yahoo.com

BIBLIOGRAPHY

1. Adams D, Athalye L, Schwimer C, Bender B. A profound case of linear epidermal nevus in a patient with epidermal nevus syndrome. *J Dermatol Case Rep*. 2011; 5: 30-33. PMID: 21894253.
2. Gon-Ados S, Minelli L, Franzon PG. Case for diagnosis. Inflammatory linear verrucous epidermal nevus. *An Bras Dermatol*. 2010; 85: 729-731. PMID: 21152805.
3. Vujevich JJ, Mancini AJ. The epidermal nevus syndromes: multisystem disorders. *J Am Acad Dermatol*. 2004; 50: 957-961. PMID: 15153903.
4. Mills O, Thomas LB. Basaloid follicular hamartoma. *Arch Pathol Lab Med*. 2010; 134: 1215-1219. PMID: 20670146.
5. Lee MW, Choi JH, Moon KC, Koh JK. Linear basaloid follicular hamartoma on the Blaschko's line of the face. *Clin Exp Dermatol*. 2005; 30: 30-34. PMID: 15663498.
6. Díaz-Martínez B, Izquierdo-Estirado M, Alramadan M, Menéndez-Ramos F, Buedo I, Arrieta-Peña E. Hamartoma folicular basaloide lineal unilateral. *Med Cutan Iber Lat Am*. 2009; 37: 251-256.
7. Brown AC, Crounse RG, Winkelmann RK. Generalized hair-follicle hamartoma, associated with alopecia, aminoacidura, and myasthenia gravis. *Arch Dermatol*. 1969; 99: 478-493. PMID: 4238829.
8. Delacretaz J, Balsiger F. Multiple familial follicular hamartoma (author's transl). *Dermatologica*. 1979; 159: 316-324. PMID: 478069.
9. Kato N, Ueno H, Nakamura J. Localized basaloid follicular hamartoma. *J Dermatol*. 1992; 19: 614-617. PMID: 1491090.
10. Mehregan AH, Baker S. Basaloid follicular hamartoma: three cases with localized and systematized unilateral lesions. *J Cutan Pathol*. 1985; 12: 55-65. PMID: 3973186.
11. Jimenez-Acosta FJ, Redondo E, Baez O, Hernandez B. Linear unilateral basaloid follicular hamartoma. *J Am Acad Dermatol*. 1992; 27: 316-319. PMID: 1517495.
12. Happle R, Tinschert S. Segmentally arranged basaloid follicular hamartomas with osseous, dental and cerebral anomalies: a distinct syndrome. *Acta Derm Venereol*. 2008; 88: 382-387. PMID: 18709310.
13. Itin PH. Happle-Tinschert syndrome. Segmentally arranged basaloid follicular hamartomas, linear atrophoderma with hypo- and hyperpigmentation, enamel defects, ipsilateral hypertrichosis, and skeletal and cerebral anomalies. *Dermatology*. 2009; 218: 221-225. PMID: 19005246.