

Case Report

Peri-Ocular and Facial Multiple Hereditary Infundibulocystic Basal Cell Carcinoma

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Purpose: To report two cases of Multiple hereditary infundibulocystic basal cell carcinoma (MHIBCC) in two successive generations of a family and to discuss its management.

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Design and Methodology: This an observational case report in an institutional setting, reporting on two patients of successive generations referred by the department of dermatology.

Clinical management: Both patients had cosmetically noticeable lesions for a very long time and did not look like typical basal cell carcinomas. Due to uncertain clinical diagnosis biopsies were taken and on the basis of differing results, a controversy of was stirred up. Histological slides and clinical photographs were sent for second opinion whereby, a definitive diagnosis was reached.

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Conclusions: Combination of minimally invasive surgery, cryotherapy and observation should form the basis of clinical and surgical management. This saves the patient from aggressive and unnecessary surgery that can be disfiguring. This is a diagnosis which should be kept at the back of one's mind when faced with similar lesions with a family history.

Infundibulocystic basal cell carcinoma (IBCC) is a newly described variant of basal cell carcinoma (BCC). There are only a few reports in current literature on its presentation, clinical course and management.

IBCC with follicular differentiation was first reported in 1987¹ and the term Infundibulocystic basal cell carcinoma was proposed in 1990². MHIBCC as a distinct entity was first described in 1999³. IBCC maybe found as a sporadic lesion, or as part of a hereditary syndrome with diffuse involvement and multiple tumors³.

We present two cases of MHIBCC in successive generations (mother and daughter) affecting the eyelid margins, periorbital skin and face. Both patients underwent different modalities of treatment. The importance of correct diagnosis is mentioned which would aid in reducing unnecessary surgery, which can be disfiguring in a cosmetically sensitive area.

MATERIALS AND METHODS

The patients were referred to the oculoplastic service from the dermatology department. The daughter was seen first, but she did not mention the fact that her mother also had similar lesions. The mother was seen a few weeks later and she gave a positive family history of similar lesions with similar distribution. After the diagnosis was established, informed consent for open publication was obtained from both patients.

Patient 1: A 43 yrs old woman was referred from dermatology for multiple fibro epithelial nodules and papules on the face, eyelids and back for the last 20 years. Some of these lesions were pigmented as well. No palmer pits, frontal bossing, hypertelorism, scoliosis or other extra cutaneous findings were observed. Her presumed diagnosis by the department of dermatology was sebaceous keratosis or fibroepitheloid polyps. Some of the forehead lesions were treated with liquid nitrogen with acceptable

results. We were requested to sort out her eyelid marginal and periocular lesions. When we examined the patient, it seemed that the lesions were more similar to lipoid proteinosis. Fig 1 (Patient 1 pre op), Fig 2 (Patient 1 pre op), Fig 3 (Patient 1 post op). No other ocular or systemic co-morbidity was noted except for mildly raised cholesterol for which she was being treated. Her mother is affected but 2 sisters do not have any skin lesions. She has no children. She underwent excision biopsy and Cryotherapy of some lesions. The histology report varied from BCC's to Trichoepitheliomas. The patient is under long-term review and is awaiting further treatment of lesions of concern to her.

Patient 2 was a 69 yrs old healthy female. She was also referred from dermatology department. She had multiple fibroepithelial nodules and lesions on face, eyelids and periocular skin. Many of the lesions were pigmented. There was one large lesion on right upper eyelid. Ocular examination was otherwise unremarkable. The patient had diet-controlled Type II diabetes for 2 years. She underwent shave excision/biopsy of the lesion on right eye upper lid and it was surprisingly reported as a completely excised BCC! Fig 5 (Patient 2 pre op), Fig 6 (Patient 2 post op), Fig 7 (Patient 2 post op). This stirred up controversy and therefore she underwent further multiple biopsies taken with narrow margins. Histology report varied from incompletely excised BCC's to Trichoepitheliomas.

Both sets of histology slides together with clinical photographs were sent for expert opinion and they came back with a diagnosis of multiple hereditary infundibulocystic basal cell carcinomas (MHIBCC).

DISCUSSION

BCC is the most common human malignancy which usually affects the elderly and Caucasians. The most important risk factors are fair skin, inability to tan and chronic exposure to UV radiation. 90% of lesions involve the head and neck region. Usually it is locally invasive but metastatic disease has been reported. BCC may be of various types, nodular, ulcerative,

cystic, melanotic and sclerosing (morphae). Sometimes multiple BCC may appear as part of syndromes e.g. Xeroderma pigmentosum, nevoid basal cell carcinoma syndrome.

Tozawa and Ackermann described a series of BCC's with follicular differentiation in 1987¹. They studied 15 biopsy specimens and described them as small well circumscribed lesions in superficial dermis composed of basaloid neoplastic cells associated with Infundibular cysts. Interestingly only one specimen was from an eyelid. On the basis of presence of peripherally palisading basaloid cells they concluded that these were basal cell carcinomas with follicular differentiation. This generated a controversy in literature. In 1990 Walsh and Ackerman proposed the term Infundibulocystic basal cell carcinoma on the basis of significant histological differences affecting solitary lesions only. In 1999 Requina et al³ described for the first time two families of patients with multiple Infundibulocystic BCC's as a distinct genodermatosis and they suggested the term MHIBCC.

Clinically the lesions were said to occur mainly on the face. Other reports in literature have reported infundibulocystic BCC's lesions on the trunk, extremities head and neck. It maybe found as a sporadic lesion or as a hereditary syndrome with diffuse involvement and multiple tumors. Typical presentation includes multiple pearly papules and dome shaped nodules, which may or may not be pigmented, scattered over the eyelid margins, periorbital skin, face, and the nasolabial folds. Inheritance pattern appears to be autosomal dominant.

The early stages of MHIBCC show numerous small epithelial components, which arise from previously normal hair follicles⁴. Most lesions tend to be superficial with no involvement of deep reticular dermis. Buds and cords of basaloid and squamoid cells in radial and anastomosing pattern are typical. Stroma is scant, myxoid and with a few Infundibular and cystic structures containing melanized and cornified cells maybe seen³. Absence of fibrocytes and scant stroma differentiates IBCC from trichoepithelioma, which tends to have an abundant and highly fibrous stroma with cribriform pattern^{2,3}.

Table 1: Key Histopathological differentiation among Hair Follicle Hamartoma (HFH), Trichoepithelioma (TE) and Infundibulocystic Basal Cell Carcinoma (IBCC) in Fully Developed Lesions⁶

Features	BFH	TE	IBCC
Cell	Anastomosing	Some	Basaloid cells with hyperchromatic pleomorphic nuclei. A

composition	basaloid cords and strands from lower portion of hair follicles	peripherally palisading basaloid cells.	few necrotic cells and rare mitosis. Some peripherally palisading basaloid cells. Several tiny cysts lined by follicular epithelium & some cornified cells. Melanin pigment maybe present
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Current literature is not clear on the course and management of MHIBCC. The age of presentation, number of tumors and distribution appears to vary from case to case. These lesions appear to be less aggressive than other types of BCC and they tend to remain small for a long period of time or grow very very slowly and show little tendency to ulcerate³. A non-surgical expectant management has been advocated⁵, which has been partly employed in our patients. The lesions in our patients had been there for more than 15-20 years without any change in character. Standard surgical procedure involves excision of lesions with 3mm clear margins. In MHIBCC this approach is not practical given the large number of lesions located in cosmetically and functionally sensitive areas. In our patients the lesions were removed piecemeal with enough time between surgeries to allow skin healing and new tissue growth till such a time that the surgical results were cosmetically acceptable to the patient

CONCLUSION

MHIBCC lesions can pose a difficult problem to deal with surgically and should be included in differential diagnosis of multiple BCC, and in particular ophthalmologists should be aware of this relatively newly described entity. Combination of minimally invasive surgery, cryotherapy and observation should form the basis of clinical and surgical management. This saves the patient from aggressive and unnecessary surgery that can be disfiguring. It is essential to furnish all relevant clinical history to the pathologists for accurate assessment and proper histological diagnosis. Moreover, if a shave excision biopsy specimen is reported back as a completely excised BCC, it should alert the clinician to the possibility of infundibulocystic BCC and the pathologist should be asked to review the specimen in greater detail especially looking for histological features consistent with IBCC.

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Conformity with Author Information

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REFERENCE

1. **Ackermann TT.** Basal Cell Carcinoma with follicular differentiation. *Am J Dermatopathol.* 1987; 9: 474-82
2. **Walsh N, Ackerman A.** Infundibulocystic basal cell carcinoma: a newly described variant. *Mod.Pathol.* 1990; 3: 599-608.
3. **Requena L, Farina M, Robledo M, et al.** Multiple hereditary infundibulocystic basal cell carcinoma: a genodermatosis different from nevoid basal cell carcinoma. *Arch Dermatol.* 1999; 135:1227-35.
4. **Toyoda M, Morahashi M.** Infundibulocystic Basal cell carcinoma. *Eur.J Dermatol.* 1998; 8: 51-3
5. **Herman AR, Klaus JB, Greenberg RA, et al.** Multiple infundibulocystic basal cell carcinomas: Case report of unique unilateral presentation. *Dermatol Surg.* 2003; 29: 436-9.

6. **Kelly SC, Ermolovich T, Purcell SM.** Nonsyndromic Segmental Multiple Infundibulocystic Basal Cell Carcinoma in an Adolescent Female. *Dermatol Surg.* 2006; 32: 1202-8.