BASAL CELL NEVUS SYNDROME (GORLIN'S SYNDROME) (A Case Report)

A. KAMATH,* L. MARQUIS † AND T. K. MEHTA ‡

Summary

A case of Basal Cell Nevus Syndrome-Gorlins Syndrome-is presented. Its varied clinical manifestations and multi-organ system involvement are emphasised. Our case presented primarily with cutaneous manifestations i.e. multiple basal cell epitheliomas, multiple epithelial cysts and pitting of the palms. Systemic involvement was minimal. Mesenteric cysts were present. No osseous, neurologic, ophthalmic, reproductive or other manifestations were encountered.

Introduction

Basal cell nevus syndrome is an autosomal dominant genetic disorder occurring with equal frequency in males and females. To our knowledge, it has not so far been reported from India. Case reports of probable examples of this syndrome date back to 1894. In 1932 Nomland[1] recognised the nevoid origin of the cutaneous tumours. In 1939 Straith[2] noted the association between jaw cysts and nevoid basal cell epitheliomas. In 1951 Binkley and Johnson[3] suggested a multiorgan system involvement while Howell and Caro[4] in 1966 served to focus attention on the syndrome.

Case Report

A 56 year old female patient was seen in the skin O. P. D. of the Nair Hospital, Bombay. She complained of multiple pigmented lesions on the face, scalp, neck and arms for 3 years. The initial lesion to appear was a small pigmented papule on the scalp which steadily progressed to a size of 2.5 cms. in diameter. A few months later similar lesions appeared at other sites. None of the lesions showed any tendency to subside, neither was there any exacerbation on exposure to sunlight. Her past history revealed that she had been operated 8 years earlier for a large mesenteric cyst.

Family history—Patient had an elder sister who showed no basal cell epitheliomas or any other clinical manifestations of the syndrome. Both her parents were dead and patient could not recollect either of them having had skin lesions similar to hers. Patient had no offspring as she was widowed at a very early age.

On general examination, patient was well built, well nourished and of average intelligence. The facies showed well developed supraorbital ridges with a broad nasal root. All her teeth were extracted and patient used an artificial denture. Over the abdomen a right paramedian incision scar was seen.

* Registrar
† Hon. Asst. Dermatologist and Venereologist
‡ Hon. and Chief Dermatologist and Venereologist
Nair Hospital and T.N. Medical College, Bombay.
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Local examination revealed pigmented papular lesions on the face, neck and front and back of the chest. (Fig. 1 Page No. 51). They varied in size from a few millimeters to almost a centimeter. An oval lesion on the scalp about 2.5 cms. in size showed central scarring with a pearly thread like margin—morphea-like appearance. There were multiple cystic lesions on both eyelids. Pitting on the palms was noticed (Fig. 2 Page No. 51). Systemic examination revealed no abnormal findings.

Investigations

Routine investigations were normal. X-rays of the jaws, ribs, vertebrae, hands and skull revealed no abnormalities. Parathyroid studies were not undertaken as there were no bony changes seen.

Skin biopsy—showed multiple masses of tumour cells in the dermis of varying size and shape. In the masses of tumour cells, the nuclei of the peripheral layer of cells showed palisade arrangement, whereas the nuclei of the cells towards the middle presented a haphazard arrangement; a picture diagnostic of solid basal cell epithelioma (Fig. 3 Page No. 51).

Discussion

Basal Cell Nevus Syndrome is an autosomal dominant genetic disorder. However, our case presented no pertinent family history. Basal cell epitheliomas are reported to appear early in life, usually by the third decade[5]. In our case the epitheliomatous lesions began rather late in life—in the fifth decade. Our case presented primarily with cutaneous manifestations—the facies was typical, with hypertelorism and broad nasal root multiple basal cell epitheliomas, multiple epithelial cysts over the eyelids and keratin defects over the palms were the other cutaneous abnormalities.

Basal cell epitheliomas may easily be mistaken for ordinary pigmented nevi. Besides superficiality, lack of invasiveness characterise the lesions for many years, suggesting a biologic behaviour different to that of the ordinary basal cell epithelioma[6]. However, each tumour is capable of local invasion and constitutes a threat to the patient as serious as the ordinary type of basal cell epithelioma. Maddox[7] reviewing the histopathology of the epitheliomas, divided the tumours into 5 types and estimated their frequency as follows — solid type 72%, adenoid type 27%, cystic type 19%, morphea type 17% and superficial 6%. In our case the histopathology showed solid type of basal cell epithelioma.

Epithelial cysts occur with increased frequency in this syndrome. though they are not distinctive. Areas of defective keratinization on palms and soles are distinctive and diagnostic when present.

Of the systemic involvement, osseous anomalies like jaw cysts are commonly seen. Minor rib and vertebral anomalies may also be encountered. Maddox's cases had jaw cysts and skeletal defects associated with basal cell epitheliomas. Binkley's case showed bone anomalies, dystopia canthorum and basal cell epitheliomas. Besides, neurologic, ophthalmologic, reproductive and other miscellaneous anomalies have been reported.

Conclusion

This case report serves to focus the attention on the varied manifestations of Gorlin's Syndrome, and to emphasize their diagnostic importance. In cases presenting with multiple basal cell epitheliomas a search for other co-exising anomalies is mandatory.

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Final Diagnosis : Aspergillosis

Patient used to feed pigeons often in the open spaces near his village. Retrospective review of the polyp histology revealed presence of broad septate filaments consistent with a diagnosis of Aspergilloma. Biopsy of the lesion showed a granuloma and P. A. S. positive branching septate filament. Culture grew Aspergillus species. The infection had reached a stage of local invasiveness with extensive tissue necrosis and intracranial involvement.