OSTEOPOIJKILOSIS AND CONNECTIVE TISSUE NAEVI: A SYNDROME OF HEREDITARY POLYFIBROMATOSIS*

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Touraine noted that certain states of nodular fibromatosis of the connective tissue in the skin and other organs are not only frequently associated, sometimes in recognizable syndromes, but are also inherited in simple dominance. The complex of hereditary polyfibromatosis includes fibromas (histiocytomas) of the skin, keloids, knuckle pads, Dupuytren's contracture of the palmar fascia, Peyronie's induration of the corpus cavernosum, uterine fibroids, fibromas of the tongue and nasopharynx, solitary or multiple fibromas of the bones, osteopokilosis, multiple enchondromas, osteopetrosis, melorheostosis, osteogenic exostoses and arthritis deformans, to mention only some of the elements. Connective tissue naevi should now be added to the list.

Osteopokilosis

Osteopokilosis (osteopathia condensans disseminata; familial disseminated osteosclerosis; spotted bones) is characterized by sharply circumscribed islands of compact bone (1 - 10 mm diameter) in the spongiosa of the bones. Lesions are usually multiple and symmetrically distributed and consist of numerous regularly arranged trabeculae of varying thickness arranged, for the most part, in the axis of the bone. They are most closely populated in the epiphyses and metaphyses of long bones, and in the pelvis, hands and feet, but they may be found in any bone; the cranial bones are rarely affected. Osteopokilosis is a benign and usually symptomless condition that is commonly ('always', said Fairbanks in 1951) discovered by chance in the course of a radiological examination for some unrelated disorder. It is purposefully sought by those aware that it is an element of polyfibromatosis. It may be an isolated element or be associated with other signs such as Peyronie's disease, dermatofibrosis lenticularis disseminata (Buschke-Ollendorff syndrome), striae atrophicae, keloids, scleroderma or sclerodermiform lesions, keratoma palmoplantare dissipatum (Aigner syndrome), keratoderma palmoplantare diffusum or connective tissue naevi. Such associations are discussed by Touraine and by Pastinszky and Csató.

Connective Tissue Naevi

Hamartomas of the dermal connective tissue have been described under a great variety of titles, of which the most important will be noted later. Although it is possible to make a rough classification of such tumours on clinical grounds, most authorities confess their inability to distinguish one from the other histologically and their group title of connective tissue naevi is the most convenient for the present time.

Both Rook et al. and Cramer and Kahler divide the group into 3 types:

1. Shagreen skin: An oval plaque (up to 10 cm x 5 cm) of papulated or crinkled skin that may be yellowish, brown or normal in colour and is usually found in the lumbo dorsal area, and occasionally elsewhere on the trunk. This naevus may be a monosymptomatic variety of the tuberous sclerosis (epiloia) syndrome of which it is often a component.

2. Naevus elasticus regionis mammariae. Cramer and Kahler are dubious about the validity of this title for a condition which is identical with that described by Lipshütz as cobblestone connective tissue naevus, and name it 'so-called' naevus elasticus. The lesions are yellowish or skin-coloured papules, sometimes perifollicular, in patches which may be quite extensive on the chest or back. This type would include cases called 'collagen naevi'.

3. Nodular forms: large nodular disseminated naevi. The lesions are smooth, yellowish or skin-coloured, slightly elevated nodules that may be discrete or aggregated into small or large plaques. They are commonly found on the thighs, buttocks and lower abdomen, and sometimes on the shoulder girdle and arms. Cases of juvenile elastoma and naevus elasticus en tumeurs disséminées fall in this category.

Histological reports on such naevi usually tell of coarse, thick collagen bundles and of elastic tissue that may be normal, reduced in quantity or hypertrophic, but Montgomery considers that as different authors are liable to make different interpretations of such changes it is best to collect them all under the heading of connective tissue naevi. Cramer and Kahler concur, saying that differentiation on histological grounds using current techniques is impossible, and that on casual examination the sections look remarkably like normal skin.

Calnan dissents and would retain the title of juvenile elastoma for cases of type 3 (the type usually found with osteopokilosis) which he considers histologically and clinically different from what he calls 'cobblestone' or 'collagen naevi'.

Familial Occurrence

Osteopokilosis has been observed in 8 members of a family in 3 generations by Hinson who cites other such cases—Verhoeve (father, son and daughter) and Wilcox (father and daughter). Busch found 14 cases of osteopokilosis in 3 generations in a family of whom he examined 41 members; 6 of the 14 also had dermatofibrosis lenticularis disseminata, but skin lesions were not found as an isolated phenomenon. Berlin et al. found 8 cases of osteopokilosis in 3 generations (56 people examined); 6 of the 8 had dermatofibromas and 2 others had skin lesions alone. In another family they found 3 cases of osteopokilosis in 2 generations (37 people examined); one had dermatofibromas, another an exostosis on a toe phalanx.

Familial occurrence of osteopokilosis and connective tissue naevi of the large nodular disseminated type was noted by Smith and Waisman (father and son; daughter had naevi only), Cairns (father and daughter) and Harman (brother and sister).

Pautrier and Woringer found large nodular connective tissue naevi in mother, daughter and niece. Prof. A.

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Basset of Strasbourg was good enough to retrace these cases and persuaded the mother to have a radiological examination which revealed lesions of osteopoikilosis (Fig. 1).

![Fig. 1. Osteopoikilosis found by Basset on re-examining Pautrier and Woringer's case of connective tissue naevi.](image)

The list is now augmented by the cases in mother and son described below.

**CASE REPORTS**

**Case 1**

A White woman, aged 32 years, had in early childhood developed a crop of flat, yellowish lumps in the skin of the lower abdominal wall. These disappeared when she was about 15 years old and at about the same time other lesions appeared on the thighs and elsewhere, and persisted unchanged and causing no symptoms.

On both outer thighs were irregular oval plaques (10 cm × 6 cm) elevated 2-3 mm above the surface and covered by skin of normal colour and appearance. The lesions were firm and rubbery in consistency and attached to skin but freely movable on the subcutaneous fat. A few small, round lesions (0.5-1 cm diameter) were present on the trunk, arms and legs. The larger lesions were clinically typical of connective tissue naevi of the large nodular disseminated type. Some ugly keloids had followed vaccination on the left upper arm and minor pustules on the chest. Because of these she refused biopsy.

Reminded by Dr E. Swart, who had recently seen such a case demonstrated by Calnan in London, I looked for and discovered osteopoikilosis (Fig. 2).

**Case 2**

A White boy, aged 6 years, son of case 1, presented lesions of the skin of the lower abdominal wall. The lesions were multiple, round (about 2 cm diameter), slightly elevated, rubbery plaques attached to skin that was normal except for a yellowish-white tinge. There were no other cutaneous abnormalities apart from pityriasis alba of the face. Radiological examination showed osteopoikilosis.

The boy's only sister and his grandparents were said to have no such skin lesions or any other sign of polyfibromatosis.

**DISCUSSION**

The discovery, in yet another instance, of osteopoikilosis and large nodular disseminated connective tissue naevi in 2 generations suggests that the syndrome deserves a place apart in the complex of hereditary polyfibromatosis. The question is: what place?

Smith and Waisman and Danielsen et al. have remarked on the similarity of this syndrome to that described by Buschke and Ollendorff. Judging from the description of the lesions of dermatofibrosis lenticularis disseminata given by Buschke and Ollendorff and by others, I would go further and say that the 2 syndromes are identical.

Some cases reported under the title of Buschke-Ollendorff syndrome do not conform to the original description. An example is that of Pastinszky and Csató where the lenticular dermatofibromas are clearly of the kind thus...
I have thought long and hard about a suitable topic for this, my valedictory address. Those of you who have had anything to do with me on or off committees will, I hope, acknowledge that my 'theme song' has always been "negotiate, talk to the other fellow, don't shoot at him... try to establish dialogue, communicate if you can". I did seriously consider as my subject 'communication'. In our modern world this offers numerous fascinating and diverse aspects, but I was forced to the certain conclusion that it would require an intellectual capacity greater than mine to do justice to such a subtle, philosophical concept.

While I was languishing in a sickbed, in the hands of the physicians, some months back, I conceived a much more mundane and topical subject which has necessarily interested and concerned me in the past year, and which urgently needs a great deal of new and enlightened thinking. I refer to our hospital and medical systems. Does anyone think that they are efficient and workable? I certainly do not. I doubt whether they are best for both patient and doctor? I intend to compare for you, as briefly and simply as I can, the current provincial hospital regimes in South Africa, and then to compare and contrast our hospital and medical schemes with those in other countries. To gather this information in the time available has been a fair task and there are undoubtedly some inaccuracies and inadequacies, which I trust do not detract seriously from the theme. I believe this to be the sort of study which could, and should, be further pursued by the right sort of select committee, not only within the Association, but at provincial and national levels.

Every province in South Africa has its own scale of hospital fees and its own hospital Ordinances. The only common meeting point is at the Administrators' Co-ordinating Council and through the Public Services Commission where such matters as salaries for all hospital personnel are co-ordinated, but there is a big difference in the actual administration of the hospitals in each province. There is, for instance, in the Cape an over-all charge, inclusive of all ancillary services and medicines up to a present maximum of R6.00 per day, and no charge at all for medical services rendered; in addition there is an initial booking charge on admission. Natal at present has abolished this charge up to a maximum of R5.50 in a ward and R7.00 in a private ward, to which is added the cost of all ancillary services and medicines. The Transvaal has a different schedule of charges according to computed income levels. I have not sufficiently studied the Orange Free State system to make comment. Further, in the Cape and Transvaal all patients, irrespective of whether they belong to a medical aid society or not, have their incomes computed and are charged accordingly, whereas in Natal all medical aid society patients and patients with an income over a level of approximately R1 800 per annum in the case of a single person, i.e. R1 500 for two persons, are at present classed as private patients, and precluded from outpatient treatment or admission to a provincial hospital as hospital patients. Charges are made by the Natal Provincial Administration for medical services rendered to hospital, for emergency and super-specialty treatments which cannot be given elsewhere. No charge is made for medical services in other provinces.

Here is a little simple statistical information about incomes, gleaned from Professor Watts, Director of the Institute of Social Research, University of Natal. It would unfortunately become too complicated and take too long to consider all these aspects as they relate to Whites and non-Whites, so that these figures are for Whites only and are derived from the 1966 census.

The 'modal' family income for the 10 principal urban areas, i.e. the average excluding the very wealthy, is R250 - R333 per month, i.e. R3 000 - R4 000 per year. Surprisingly, nearly 50% of White families consist of only man and wife or of a couple plus one child. Only approximately 25% of families have 3 or more children. With this basic knowledge let us consider an example of hospital costs as they may affect the most common individual case. In Natal and the Cape any couple with this average income group with one child will pay full private rates. In the Transvaal they will pay R5.50 (wardbed) or R7.00 (private ward), plus extras, in Natal. In the Transvaal they will pay R4.00 to R5.00 per day plus extras. The over-all cost is highest in Natal for the majority of income groups, and the cost in any event in all the provinces is beyond the means of the average family should they have an expensive and prolonged illness, even if they are covered by our present medical aid type of insurance. Thus 10 days in a hospital at these rates must cost R50.00 to R100.00, i.e. one-fifth to one-third of an average monthly income. This relates to admission to a provincial hospital, and much more is paid for a hospital stay. There are, in fact, many people who really cannot afford to belong to medical aid societies as they now exist, in that, if they exceed their benefits for a year, they cannot hope to pay the excess, and many of them cannot even afford the 10 - 25% of the