Brown Spots

To physicians other than dermatologists the distinction between the freckle and the lentigo may seem to be at best of academic interest. Yet these two small brown spots are fundamentally different in structure, natural history, and associations. The freckle is induced by light in susceptible people and may fade if exposure to light is reduced. The melanocytes which form it are abnormally large. The lentigo is not influenced by light, tends to persist for years or indefinitely, and is formed of an increased number of apparently normal melanocytes. The development of a few or many lentigines during childhood or early adult life, apparently randomly distributed over the body surface, is so common as to be regarded as physiological. The term lentiginosis has been applied when lentigines are present in exceptionally large numbers or in a distinctive distribution.

The distribution of lentigines in a characteristic pattern in more than one member of a family has attracted the attention of dermatologists, and A. Touraine in 1955 reviewed the scattered literature. He recognized two distinct patterns. In one of these, which he had earlier described and named centrofacial lentiginosis, the lentigines appear in early childhood in a horizontal band across the centre of the face. Associated defects include confluence of the eyebrows, a high arched palate, spina bifida, and mental retardation. Inheritance is apparently determined by an autosomal dominant gene.

The second distinctive pattern of lentiginosis occurs mainly around the orifices, and the principal associated abnormality is polyposis of the gastrointestinal tract. This syndrome, also determined by an autosomal dominant gene, was first described by J. L. A. Peutz in 1921. In 1946 Touraine and F. Couder gave a detailed account of the syndrome and collected 31 cases. H. Jeghers and his colleagues in 1949 reported further cases, but the now familiar eponym Peutz-Jeghers syndrome shows a regrettable disregard for historical precedence.

Other patterns of lentiginosis have been reported in single patients or in two or more members of a family, but in many of the reports the clinical description is incomplete, and if associated defects in other organs were sought they are not mentioned. But it is now clear that widespread lentiginosis is a feature of at least one distinctive hereditary syndrome. That syndrome, for which the term progressive cardiomyopathic lentiginosis has been proposed, was first identified by E. J. Moyer, and has recently been critically reviewed and characterized by D. E. Polani and Moyer, who have studied eight patients. Lentiginosis is present in infancy, but the number of lesions increases during childhood until many hundreds are present, irregularly scattered over the body. Hypertrophic obstructive cardiomyopathy, affecting predominantly the left side, may cause severe symptoms or be detected only on electrocardiography. Other features of the syndrome are impaired sexual development and stunted growth. In some cases there is also some degree of intellectual impairment.

The inheritance of this syndrome is determined by an autosomal dominant gene. Polani and Moyer discuss the possible relationship between the two principal effects. The cardiomyopathy seems likely to be related to the lentiginosis because melanocytes which form the latter are derived from the neural crest, which also contributes to the structure of the heart. Less plausible is the hypothesis that a disorder of inotropic amines in the lentigines gives rise to abnormal cardiac function.

The relationship of cardiomyopathic lentiginosis to other reported syndromes with profuse lentiginosis has not yet been established. For example, O. F. Hornstein and F. Weidner have recently reported a further case of the association of lentiginosis with congenital deafness, which A. J. Capute and colleagues had described in 1969. The patients described by R. J. Gorlin and colleagues showed generalized lentiginosis, electrocardiographic abnormalities, pulmonary stenosis, and deafness. Inheritance was of autosomal dominant type, with variable expressivity. Other reports present different variations, with lentiginosis, cardiac defects, and deafness as the key features. Only the careful study of further families will elucidate the genetic and pathological problems raised by this association of defects. It is evident that any person presenting either generalized, centrofacial, or peri-orificial lentiginosis deserves thorough investigation.

Phoenix from Physical Medicine

In 1931 a decision was taken to amalgamate two sections of the Royal Society of Medicine—the Section of Balneology and Climatology and the Section of Electrotherapy. The name chosen to represent the interests of members of these sections was “Physical Medicine,” and from this originated a new specialty under the same name.