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Dupuytren diathesis

Recognition of the existence and importance of the diathesis to Dupuytren's disease (DD) in all patients and their relatives, has in recent years changed the surgical advice offered. No longer is an individual finger assessed and treated — it has become clear that total assessment of the patient is imperative if proper advice is to be proffered.

Derived from the Greek root to dispose or distribute, the word *diathesis* can be seen to give a guide to the disposition of what the Oxford English Dictionary terms 'a permanent condition of the body which renders it liable to a special disease'. This permanence has been demonstrated to depend on an inherited genetic pattern or 'disposition' of the chromosomal material responsible for the development of DD. Therefore it should be clear that it is impossible for a patient with DD to have no diathesis.

It would appear that there has been some difficulty in appreciating this concept of DD, McFarlane (1985) expressed surprise when a computer study of 1000 operated patients confirmed not only the existence of a diathesis but its importance in the assessment of the natural history of DD.

Of the utmost importance in counselling patients with DD is the expected outcome of treatment. It can safely be stated that unsatisfactory results fall into two categories — early and late. The early or postoperative complications of hematoma, infection and skin necrosis are due to the surgeon. The late appearance of new Dupuytren's tissue, either in recurrence or extension, is due to the patient. In particular it is due to the strength of the diathesis of the patient to the production of this palmar fibroplastic tissue.

It is therefore manifestly important to assess as well as possible the factors indicating the strength of the diathesis, and advise the patient on the likelihood or not of further development of the fibroplastic tissue of Dupuytren's disease, either in the operatively cleared zone (recurrence) or beyond the area of operation (extension). To be precise, the radical procedure of skin replacement (dermofasciectomy) will be advised if recurrence is deemed probable, but it will not be advised if recurrence is regarded as unlikely.

What then are the features to be considered in assessing the strength of diathesis and hence the risk of recurrence in any particular patient?

RACE

Despite the extremely rare and patently incontestable reports of DD in isolated Africans (Furnas 1979; Mennen 1983), DD has not been seen in non-Europeans until the recent reports in Japanese by Egawa et al (1985). These will be placed in perspective presently.

Even within Europe there is wide variation not only in incidence but also in the strength of manifestation. Thus it is difficult — indeed impossible — to compare objectively the clinical manifestations of the condition and its treatment, even within Europe itself. The condition treated in Vienna by Millesi is clearly at a lower level of penetration in the population than that treated in Ireland by Varian. Hence the need for skin replacement, because recurrence is higher in the UK than in central Europe.

The incidence in Scandinavia, the Netherlands,

Scotland, England and Ireland is higher than in the Mediterranean basin. In Italy there is a far higher incidence in the north than below Naples; this is consistent with the introduction of genetic predisposition in the north-west from French invasions and in the north-east from Austro-Hungarian invasions. Despite the fact that Australia has the highest Greek population outside Greece, I have seen only one Greek male with very mild DD, — palmar nodule only — and on questioning he revealed that his mother was Irish!

An explanation of this north-west European dominance of DD has been that it was in the genetic spectrum of the Celts. More likely, in view of their wide raiding patterns, it could be regarded as a Viking disease. Indeed the occasion of finding DD in a Sicilian with blue eyes is likely to be due to the Norman invasion of Sicily in 1066 by the brother of William the conqueror, who invaded England in the same year.

A neat study by Brouet (1986) in Toulon has noted a striking difference in the incidence of DD in the dark-eyed Mediterraneans of local stock and that in the blue-eyed Nordic stock, comprising sailors who for generations have settled in the milder climate of this French coast and have manifestly brought their genetic pool with them. A far stronger diathesis is noted in the Toulonese of Nordic than of local Mediterranean stock.

But what of the recent reports from Japan? There is no historical evidence of Celtic or Viking invasion there. While naval visits from American and European ships may possibly have seeded a few cases, another possibility exists. It is to be noted that the diathesis of the Japanese patients is very low. Very few progress to surgery for correction of deformity. The demography of the condition in Japan has not yet been established but it is well known that the tall, paler Hokkaido population are already genetically distinguished from the shorter, darker southern Japanese. In a study of the linguistic patterns of the northern people of Europe and Asia, Collinder (1957) has demonstrated that a group of linguistically related peoples at the northern region of the Urals, before the Vikings dispersed both east and westwards along the line of the Arctic Circle, passed through Mongolia to the eastern littoral. It is certainly not impossible that this basically northern European

strain was thus introduced in a very diluted form into some areas of Japan.

The absence of any genetically pure Negro in the USA after four centuries of cohabitation makes DD in a black patient of no interest in this racial consideration. Conversely the high incidence in Canada, where kilts are commoner than in Scotland itself, supports the known Viking excursions through Iceland and Greenland to the mainland of Canada — not to mention the subsequent millennium of migration from the British Isles. The same Scottish and Irish predominance in the early white settlers of Australia explains, by this distillation of the genetic pool, the high incidence in this country and in the white settlers of New Zealand.

This racial factor is by far the most important single element determining the incidence of DD. The patient's universal enquiry: 'Why do I have this condition?' can truthfully be answered: 'You were born to get it'.

AGGRAVATING FACTORS

The strength of the diathesis can be increased by genetically related epilepsy or diabetes mellitus. In a genetically predisposed population other factors seem to dispose the clinical manifestation of the condition to appear earlier or more commonly than in their absence. These factors include chronic pulmonary disease, cirrhosis and alcoholism, all of which have classically been associated with other changes in the connective tissues of the hands, such as clubbing of the fingers and 'liver palms'. The difficulty in grasping the concept of a diathesis has been failure to realize that all these predisposed populations have a diathesis of some degree, but the clinical manifestation may remain latent. It is only in the presence of some added factor that DD may appear or progress in these special disease groups.

The simplest of these factors is disuse. The onset or aggravation of DD in patients rendered inactive by a major injury, such as major fracture of the legs or back, has been seen to be followed within weeks by the onset or progress of DD often bilaterally.

Injury

In the absence of a racial predisposition to Dupuytren's disease, any amount of injury to the hand will fail to produce DD. However, where the diathesis is racially disposed the appearance or progression of DD has been noted to follow almost any injury to the limb associated with a period of swelling and disuse of the hand. These injuries range from radical mastectomy through proximal limb ruptures, dislocations of shoulder or elbow joints, forearm fractures, superficial burns of the hand, to direct local tissue disruption with hematoma or open wounds and fractures in the hand itself. That all these different identities will act through some final common pathway on the palmar connective tissue between the dermis and the palmar aponeurosis, without necessarily injuring this well defined connective tissue compartment, indicates that a basic balance of tissue turnover or entropy has been disturbed in this compartment. Whatever the basic mechanism of the metabolic change — and changes in the fat as a possible mediating element have been identified (Rabinowitz et al 1983) — it is of paramount importance to note that the palm itself does not have to be the site of the injury. Plewes' (1956) observed that every patient with Sudek's atrophy showed the palmar proliferation of DD, despite a multiplicity of causes of the Sudek's atrophy. This points to the importance of a change in the general vasomotor status of the hand as a pathogenetic pathway.

The persistent proposal of partisan theories based on rupture of the longitudinal fibres of the palmar aponeurosis fails to explain the phenomenon of onset without local injury apart from disuse and local oedema. The evidence supporting the extra-aponeurotic origin of the primary Dupuytren's nodule — apart from the observation that the primary nodule is on the palmar and never the dorsal aspect of the palmar aponeurosis — has been confirmed by the author's observation of retraction induced in the digital extensor apparatus by a dorsal plaque of DD (Hueston 1982a). Iselin (personal communication) noted a similar phenomenon. Finally, the hypothesis of pathogenesis with intrinsic rupture of longitudinal aponeurotic fibres fails to explain the phenomenon of recurrence.

Every fasciectomy must be recognized as a specific local injury to the volar compartments of the palm and fingers. It is biologically inevitable that swelling and disuse, however temporary, will follow this surgical injury. The longitudinal fibres still cited by some reactionaries as the site of origin of DD are no longer present after fasciectomy. Yet the incidence of recurrence within the operative area is now recognized as being between 40 and 60% in an Anglo-Saxon community (Hueston 1963; Rank & Chang 1978 Tonkin et al 1984; Niebauer; personal communication). The tissue where the recurrent tissue forms — very frequently within a few weeks — is the tissue of the replaced flaps of volar skin elevated for exposure. This is the tissue in which the original change leading to the primary nodule had arisen, possibly aggravated by the vasodilatation (Bauer et al 1985) and oedema (Plewes 1956) of a specific episode of regional injury and disuse (Hueston 1982b). Only by the sacrifice of the subcutaneous tissue and its overlying skin, along with the fascia, has it been found that recurrence can be avoided. This locally more radical procedure is usually reserved for the digits rather than the palm.

Occupational factors

In the absence of a specific injury to the hand or the proximal limb leading to swelling and temporary disuse, there is no evidence that manual labour initiates or aggravates DD (see Chapters 19, 24 and 25). Indeed the opposite is the case; progress of the disease is often observed when the maximum physiological use of the hand in heavy labour is replaced by a more sedentary job involving supervising or writing with, of course, the ulnar digits flexed and unused.

That the diathesis over-rules any claimed local hand activity is confirmed by:

1. The fact that there is no relation between the disease and handedness.
2. The fact that the workers are often aged 50 or older when DD is first noticed; this is normal in the natural history of the disease.
3. There may be racial groups in a job, such as Dutch or German stock in a brewery business or migrant labourers from Scotland

and Ireland in Australia. Vibrating tools have no relation to DD (Hunter et al 1944).

There is no doubt that most work in Third World nations is heavily manual and yet DD is not seen there, for the simple reason that no diathesis exists in the absence of an inherited genetic predisposition.

CLINICAL ASSESSMENT

The first clinical examination of every patient with DD must involve removal of the patient's shoes. The author's practice is to do this before noting the details of the hands. The clinician must record whether one or both soles has nodules and how extensive they are. This observation is checked at subsequent annual visits.

The presence and distribution of knuckle pads over the proximal interphalangeal joints is also noted. Knuckle pads may also be over the metacarpophalangeal joint or, most commonly, on the thumb at the interphalangeal joint. It should be recognized that depression over the joint as a result of early adhesion of the dermis to the paratenon may be present before the much commoner single or multiple nodular mass is seen (Hueston 1984). Regular annual recording will often show that these knuckle pads occur on new digits or, rarely the knuckle pads may be noted to shrink or disappear from one or more digits. The palm may appear clear of nodules or bands in a patient with knuckle pads but it is inevitable that palmar lesions will ultimately appear.

The justification for taking these ectopic deposits of identical fibroplastic tissue masses as an index of the strength of the diathesis is that a far higher incidence of recurrence has been found in patients with knuckle pads. What is more, those patients with knuckle pads are generally younger than those without, so it is important to identify young patients with knuckle pads as having a stronger diathesis and greater likelihood of recurrence than those patients who are elderly when the condition appears and in whom no knuckle pads

are present. In such elderly patients recurrence is unlikely and less radical surgery can usually be advised.

The mode and rate of onset of the palmar changes may indicate a stronger diathesis. If the disease is rapid in onset and progress and particularly if it follows soon after a specific local injury or previous surgery, the risk of recurrence after further surgery is greater; it is inevitable if the patient is young and has ectopic deposits.

Skin involvement is difficult to assess as a prognostic factor in diathesis but if it is extensive then the field of palmar extra-aponeurotic fibroplasia is manifestly more widespread and reflects a greater tendency to the production of this tissue before — and therefore very likely after — the injury of fasciectomy.

Previous surgery should always be presumed to have been well done. Derogatory statements that the recurrence is due to the previous surgeon's poor technique show an ignorance of the fact that it is the patient who is producing the recurrence because of the strength of the diathesis. Further surgery must be directed towards prevention of recurrence by replacing the skin, otherwise the early appearance of a further recurrence will leave the critical surgeon red-faced.

Of least practical importance in assessing the strength of the diathesis is the absence of a family history. While a positive family history of one or several close blood relations with DD supports the assessment of a stronger diathesis, the absence of a family history should not lead the surgeon into presuming a low penetration of the disposition. The classical study of Ling (1963) confirmed what we find very often, namely that the patient who on the first visit denied any family history later reveals one or more relatives involved. A negative family history is the most unreliable of all the factors outlined above in the assessment of the strength of the diathesis.

The strength of the diathesis determines the treatment. Attempts to grade numerically the degree of diathesis have not yet appeared in the literature, but should be regarded with a healthy scepticism in this essentially clinical process of individual assessment.

Discussion

John Hueston has provided an eloquent account of his original concept of Dupuytren's diathesis. I would, however, take issue with his closing comment that attempts to grade numerically the degree of diathesis should be regarded with scepticism. Lord Kelvin said:

When you can measure what you are talking about and express it in numbers, you should know something about it; but when you cannot measure it, when you cannot express it in numbers, your knowledge is of a meagre and unsatisfactory kind: it may be the beginning of knowledge, but you have scarcely, in your thoughts, advanced to the stage of a science.

The goal of the statistical analysis reported here was to determine which factors, if any, contributed to an increased incidence of recurrence or extension of disease following appropriate operation; that is, to show that under certain circumstances

the disease is more aggressive and to evaluate the causative factors. Five factors were found to affect appreciably the course of the disease but, as shown in Figure 22.1 only two of these factors, 'other areas involved' and 'early onset of disease', had a statistically significant effect. Clinically this would mean that either factor, when present alone, should alert the surgeon that there was a very good chance of recurrence and extension of disease and that more extensive surgery such as extensive fasciectomy or dermofasciectomy was indicated. The other factors, 'bilateral disease', 'family history' or 'more than two rays involved', acting alone would not have an effect upon the course of the disease. However, the presence of two or three factors would probably cause aggressive disease. As shown in Figure 22.1, when all five factors are present there is a profound difference in the rate of recurrence and extension. These interrelation-

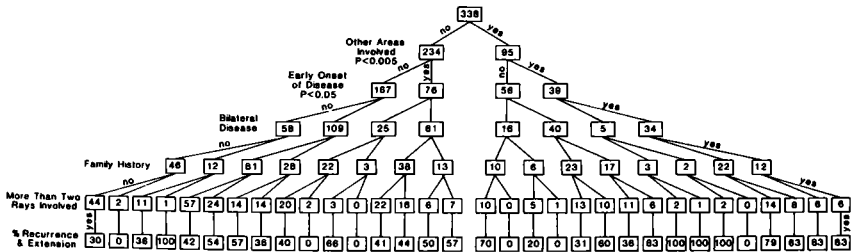


Fig. 22.1 Rate of recurrence and extension of disease according to the factors contributing to an increased diathesis. The group consists of 338 patients who had had no previous operation and who were examined 2 years or more after operation. If all five factors are present the rate of recurrence or extension is 83%. If none of the factors are present the rate is 30%. Acting alone, only the factors 'other areas involved' and 'early onset of disease' have a statistically significant effect upon recurrence and extension. Acting in combination, these five factors have a profound effect.

Table 22.1 Knuckle pads and plantar nodules: 50 Patients; 67 Hands

Family origin		Sex		Hand dominance		Hand involved		Occupation	
Northern European	92%	Male	84%	Right	86%	Right	10%	Manual	49%
		Female	16%	Left	14%	Left	10%	Non-manual	51%
		Other areas involved		100%		Both		80%	
		Family history		46%		Associated diseases		Age at onset (years)	
		Previous operation		46%		Epilepsy	16%	Male	40.2 ± 13.0
						Diabetes	6%	Female	46.4 ± 13.5
						Alcoholism	18%	Age at operation (years)	
						Trauma	18%	Male	53.5 ± 10.5
								Female	58.5 ± 12.2
Operation profile									
Hand profile				Palm	Little finger	Thumb	Anaesthesia		
Palm only	3%	<i>Operation</i>							
No palm	3%	Local		8%	8%	6%	Local	2%	
		Regional		60%	32%	65%	Regional	45%	
One ray	33%	Extensive		32%	59%	29%	General	53%	
Two rays	22%	Amputation			1%				
Three or more rays	42%								
		<i>Incision</i>					Procedure at PIP joint		
Thumb and thumb web	40%	Longitudinal		76%	99%	100%	19%		
Index finger	36%	Transverse		24%	1%				
Middle finger	37%						Complications		
Ring finger	52%	<i>Closure</i>					24%		
Little finger	66%	Suture		70%	83%	76%	Therapy		
		Open		14%	0%	0%	79%		
		Graft		16%	17%	24%	Splinting		
							42%		

ships and trends are illustrated in Figure 22.2. The percentage of hands involved increases directly with the number of rays involved for factors associated with aggressive disease.

Because 'other areas involved' is such a strong diathesis factor, a patient profile was prepared and is shown in Table 22.1. Knuckle pads occurred in 20% of surgical patients and plantar nodules in 7%. Only 50 patients (4%) had both knuckle pads and plantar nodules. Penile involvement was not included because this would eliminate females.

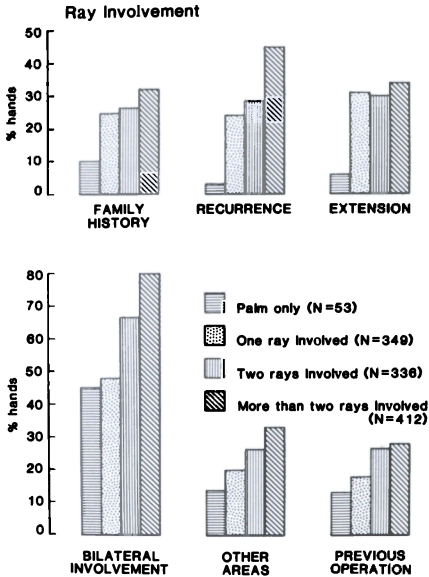


Fig. 22.2 Positive relationship and increasing trend of various diathesis factors with increasing ray involvement in the hand.

Predictably the combination of knuckle pads and plantar nodules influenced many factors in Table 22.1. Note the increased prevalence of family origin, bilateral disease, family history, previous operation, epilepsy, and alcoholism and perhaps trauma, and the marked reduction in the age at onset and operation in both sexes. There were more rays involved and more radial side disease. Skin grafts were used more frequently in the palm and fingers. There were more procedures upon the proximal interphalangeal joint and more complications, indicating that more extensive operations were performed.

Our conclusions are similar to those of Hueston but with statistical support. The most important factors are 'other areas involved' and 'early onset of disease'. These two factors stand alone and are not sex-related. Racial origin is not a strong factor simply because most of the patients studied were of northern European origin. For the same reason, bilateral involvement is not a strong factor because most patients have bilateral disease. Family history would be a significant factor if a 'proper' family history was obtained. However from a practical point of view only 25–30% patients seen by a surgeon will know of a close relative having the disease. This information is of no value in predicting the course of disease. Epileptics and alcoholics have severe and aggressive disease. It is not known whether this is genetic or drug-induced. Diabetics have mild disease; in fact, the majority do not require treatment. Concerning trauma the data are conflicting because the disease appears in younger males. However the disease is less extensive and less aggressive so trauma would not appear to be a diathesis factor.

The term 'Dupuytren's diathesis' has been in common usage for many years without being clearly understood. We have presented here some scientific evidence in support of this concept.