Chromosome 21 and Down's syndrome

Although Down's syndrome may involve trisomy of the whole of chromosome 21, the same clinical picture is produced by triplication of a minimal, critical segment at the end of the long arm. In this region, 21q22.3, lie genes for collagen type 6,[18] which may be responsible for the unusual skin texture, joint hypermobility, and tendency to perforating collagenoses in Down's syndrome.

X chromosome

Several rare genodermatoses are X-linked. The best known is hypohidrotic ectodermal dysplasia, mapped to Xq12-q13.1. Steroid sulphatase deficiency due to deletions at Xp22.32 produces a characteristic ichthyosis in affected boys. This condition may in future be correctable by gene therapy. Already the missing gene has been replaced in cultured keratinocytes from a patient with X-linked ichthyosis,[19] heralding a new era in dermatology.

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SKIN AND THE PSYCHE*

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Emotional and psychological disturbances underlie many problems in dermatological practice. Patients may be divided into four groups.

1. Those with delusions of parasitosis, depression and body image disturbances, obsessional neuroses, dermatitis artefacta and trichotillomania.

2. Dermatological conditions which may be initiated or exacerbated by stress, eg urticaria, atopic eczema, acne vulgaris, rosacea and alopecia areata. Others which may occur in association with emotional or psychiatric disturbances, eg neurodermatitis, pruritus ani, pruritus vulvae, hyperhidrosis and generalised pruritus.

3. Reactive depression and/or anxiety associated wtih their skin disease.

4. Skin disease induced by psychotropic drugs, eg lithium induced acne or psoriasis.

This short review concentrates on patients in the first group.

BODY IMAGE AND ITS DISTURBANCES

A major component of the perceived body image is cutaneous, and some areas are more important than others; the face, especially the nose, the hair and genital are all crucial in body image perception. What is desirable is continuously changing. Thus, the voluptuous woman painted as the role model by artists of the 17th, 18th and 19th centuries has been replaced in the latter part of this century by the Barbi doll. The sophisticated women in the 1990s is expected to have plenty of hair on her head, but virtually none in secondary sexual areas, such as the axillae. The pubic hair must be as inconspicuous as possible, and no hair is permissible on the arms, legs, face or chest. Breasts are expected to be relatively inconspicuous, and the ideal distribution of fat is conceptualised as prepubertal. Indeed, the body structure of a Barbi doll, if attained by an adult female, would be incompatible with normal menstruation. In addition to this almost complete negation of secondary sexual characteristics in adult women, the skin itself is expected to be like that of a baby and should therefore be wrinkle, spot and grease free. Without such a skin, many women become unhappy and their self-esteem and confidence falls, resulting in secondary depression.

The personality most vulnerable in this regard is found in those females who have never communicated well with their fellows, men or women, and in addition show narcissistic, ruminant, obsessional and perfectionist traits which render them incapable of accepting anything less than perfection. Many such individuals have a borderline personality disorder.

It should be remembered that, whilst a thin, cachectic woman may be perceived as desirable in the Western world, in societies elsewhere more traditional values prevail. Indeed, some societies see an excessively thin woman as

*Based upon a lecture delivered at the Symposium on *Dermatology* in the College on 4 May 1994.

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potentially sickly and to be avoided at all costs as far as, for instance, marriage is concerned.

Whilst the cosmetic industry exhorts women to search for younger skin, at the same time cosmetic advertising encourages women to sunbathe. A good suntan is equated with good health and beauty. Thus a conflicting message is sent. On the one hand they are told to strive for a childlike skin, on the other they are urged to photo-age their skin each summer. However, the pendulum is swinging and a pale skin is becoming more fashionable, though by no means *de rigeur* in 1994.

The concept of what is a desirable body image is also shifting in men. Whilst tattoos are becoming more fashionable for women, they are becoming undesirable in men, who may be desperate for their removal. Moreover, chest hair in men is also occasioning some distress. A shiny, oiled chest free of all hair is seen by some cosmetically vulnerable males as the target compatible with a happy life. The recent fad for the use of androgens in young men to promote muscle growth is also related to shifting body image values in males.

DERMATOLOGICAL DELUSIONAL DISEASE

Dysmorphobia or 'dermatological non-disease' describes patients rich in symptoms, especially in cutaneous areas important in body image, but poor in signs of organic disease.[1] Patients presenting in this way are ill, but the commonest psychiatric disease present is depression.[2] The terms dysmorphobia and dermatological non-disease do not constitute a final diagnosis, but a starting point for accurate psychiatric diagnosis. Patients with dermatological non-disease present with symptoms referable to three main body areas, the face, the scalp and the genital area.

Facial symptoms include complaints of excessive redness, blushing, burning feelings rather than itching, scarring, large pores, excessive facial hair and excessive facial greasiness. Symptoms may appear after or during severe emotional problems, especially where there is marital disharmony. Scalp symptoms include a feeling of intense burning, unremitting by day or night, and complaint of excessive hair loss.

Genital symptoms in males include complaints of an excessively red scrotum, discomfort in the genital area which may be described as spreading on to the anterior thighs making wearing of clothes uncomfortable, the presence of a urethral discharge and a belief of having recurrent herpes or AIDS. The female equivalent is vulvodynia (the burning vulva syndrome), where the discomfort may be so severe that the patient feels unable to sit or to lie in bed. Patients with facial and scalp symptoms are more often female, whilst those with perineal symptoms are more likely to be male than female.

Pathogenesis

This is unknown but like patients with delusions of parasitosis (see below), affected individuals are often solitary, unmarried or divorced. They usually socialise poorly and seem afraid of contact with other people. Genital symptoms in men may follow imagined or real sexual exposure. An obsessional premorbid personality is not unusual, and the commonest associated psychiatric illness is depression. It is unusual, however, for patients to admit to depression, although many do admit to early morning wakening.

In a young adult or teenager schizophrenia or a personality disorder may present with dermatological non-disease, whilst in an elderly patient dementia should be considered. There is usually no evidence of any underlying organic dermatological or neurological disease.

Other clinical features. Patients are 'doctor shoppers' and never respond to placebos. Indeed, they may develop a nocebo reaction, ie when given a placebo develop symptoms which they attribute to the agent. Consultations with dermatological non-disease patients always occupy more time than the conventional consultations with patients with organic skin disease.

Management. This is always difficult. Some patients with over-valued ideas may be amenable to superficial psychotherapy, but the patient with a true delusion, even though clinically depressed and scoring highly on depression inventories, may respond poorly to antidepressants. Usually, however, the sleep pattern improves, whilst the delusional illness persists. If a patient has a pure monosymptomatic hypochondriacal psychosis,[3] it is claimed pimozide may be helpful but the writer has had little success with this drug in this condition.

DELUSIONS OF PARASITOSIS[4]

Patients with delusions of parasitosis have an unshakable conviction that they are infested by parasites. These delusions are three times as common in women as in men over the age of 50, but affect both sexes equally below this age. In young adults the symptoms may suggest illicit exposure to drugs such as cocaine and amphetamine, but also they may be part of a shared delusions with another member of the family (*folie à deux*). Most patients are intelligent and are often thought of as being eccentric by their peers. It is often difficult to decide where eccentricity ends and madness begins.[4] Like patients with dermatological non-disease, those with delusions of parasitosis are often lonely, solitary individuals living on their own. An obsessional, premorbid personality may also be seen. The profession are well represented as patients have included doctors and even psychiatrists.

Delusions of parasitosis are sometimes seen in patients following a cerebrovascular accident affecting the nondominant hemisphere. It has also been described in patients with pellagra, vitamin B12 deficiency and severe renal disease. Lyell has stressed the importance of excluding the presence of parasites, as 6 of the patients in his series had developed the delusions following preceding real infestation.[4]

Clinical features. These are striking and the patients usually present to their medical practitioners with a small container, such as a match box, alleged to contain the insects or parasites which they had removed from their skin. More or less unintelligible diagrams may be produced. Sometimes the skin has been excoriated or treated by red hot needles or wire in an attempt to remove the parasites. Sometimes the patients are exhausted because of constant washing and cleaning rituals in their quest to rid themselves and their environment of the parasites. University zoology departments, entomologists, the British Museum, and Rentokil may all have been consulted by the almost desperate patient. Indeed, it is likely that patients with this disorder contact these agencies more commonly than they contact medical practitioners. Sometimes a general practitioner has colluded with the delusion.

Management. Patients often respond dramatically to treatment with pimozide. The drug is given in an initial dose of 2 mg, increased as necessary by weekly or twice weekly increments to a maximum, of 12 mg daily. At higher doses patients readily develop extrapyramidal symptoms and there is a risk of ventricular arrhythmias, so an ECG is important before and during therapy at these doses.[5] The drug should not be given to any patient with a prolonged QT interval or to those with a history of cardiac arrhythmia. Hypokalaemia may predispose to the cardiotoxic effects of pimozide, and care should be taken in patients with hepatic or renal dysfunction. Whilst many patients respond to pimozide, some are depressed, and are then best treated with a conventional antidepressant, sometimes in conjunction with pimozide. Patient compliance is poor and it is usual for them to defect from follow-up. In exceptional circumstances depot neuroleptics can be used.

DERMATOLOGY AND PHOBIAS

Hand eczema, particularly if it is refractory, may have been induced by repeated hand washing. A psychiatric diagnosis of obsessional neurosis may be appropriate in such instances. Patients are also encountered who are phobic about warts or pigmented moles. The latter has become more common, following publicity campaigns aimed at early diagnosis of malignant melanoma. Affected individuals, usually a mother, consult dermatologists repeatedly about their moles, and also about their family members' moles, especially on their children. Sunny holidays become banned and great stresses build up within the family. A mother with mole phobia may push for her children to have the moles removed surgically or by laser therapy. A clinical psychologist can help by giving these obsessional individuals some sort of strategy to cope with their phobic crises.

Anorexia nervosa can be regarded as a phobia about body weight and has many dermatological accompaniments such as perniosis, lanugo body hair, diffuse nonscarring alopecia, and even calluses on the fingers induced by repetitive insertion into the mouth to induce vomiting. Yellow discolouration of the skin may also be seen due to an increased blood carotene level.[6]

Erythrophobia is quite common in teenagers and young adults of both sexes. These perfectionist, narcissistic, obsessional, ruminating individuals become extremely introspective about the colour of their face. Once more a clinical psychologist or even hypnosis can help in managing this clinical situation, which does not usually respond to drug therapy.

ARTEFACT DERMATITIS[7,8]

It is often difficult for doctors to realise that a patient may, consciously or subconsciously, set out to deceive them. Artefact dermatitis is a case in point and there are several pointers to the diagnosis. The condition is more common in women than in men, and especially in those who have been employed in the health caring professions. The clinical range is wide and extends from nail biting at the one end to suicide at the other. Most patients presenting to dermatologists lie somewhere between those two extremes. The majority are teenagers or young adults with an immature personality.

Clinical features which should lead a dermatologist to a suspicion of artefact dermatitis include age, female sex, a health work background, a 'hollow' history, in that the patient is unable to give any account of how the lesions developed and bizarre lesions with linear or circular configurations. The *belle indifference*, although sometimes present, may not be well marked. Most patients readily agree to come into hospital and are happy to be the centre of attention as at clinical demonstrations to medical students and other doctors. They agree readily to photography. Vasculitis, especially on the legs, may be simulated closely, and subvariants of this condition include factitious cheilitis and psychogenic purpura.

Thick hospital records are a characteristic of the condition in patients with long-established problems, and there may have been episodes of other simulated disease. Thus Munchausen's disease by proxy (Meadow's Syndrome)[9] can have dermatological presentations, including dermatitis simulata, where the mothers, often former nurses, apply dye stuffs to the skin to try to simulate skin disease. Thick records applying to a child, where no firm diagnosis has been made and where the history is bizarre, should suggest this diagnosis.

DERMATOLOGICAL PATHOMIMICRY[7]

Recurrences of exacerbations of existing skin disease may be induced by patients by deliberate contact with agents which the patients know will damage the skin. Examples of this are contact dermatitis induced by hair dye by women who are well aware of this allergy. Millard described a group of 13 such patients, and felt that this disorder should be regarded as an entity distinct from patients who have dermatitis artefacta. He concluded that direct confrontation, without any hint of reproach, was helpful in management, in contrast to dermatitis artefacta, where it is usually unhelpful.

DERMATOLOGICAL SUICIDE

Patients presenting to dermatologists may have emotional, psychological or psychiatric problems profound enough to make them suicidal, or even to commit suicide. A retrospective study has analysed 13 dermatological patients who were known to have committed suicide in the Leeds region over the last 20 years. The largest group were those with dermatological non-disease and associated depressive illness. Females with facial symptomatology were at particular risk of suicide. Males with severe facial acne scarring were the second largest group and it should be recognised that patients with severe skin pathology, especially with scarring, of important body image areas such as the face are at high risk of reactive depression and suicide. The third group were patients with longstanding, severe and debilitating organic skin disease, such as systemic sclerosis. Severe reactive depression may develop in this clinical situation and be followed by suicide.

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