Down's syndrome and the general practitioner

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SUMMARY. People with Down's syndrome form a heterogeneous group sharing a single constant feature — an extra chromosome. This paper reviews the many clinical problems associated with Down's syndrome and emphasizes the prevention of secondary handicapping conditions. Current policies on antenatal screening for Down's syndrome are discussed. The review draws attention to the need for general practitioners to see themselves as part of a network of community services providing support to people with Down's syndrome and their families.

Introduction

DOWN'S syndrome is the most frequently observed chromosomal abnormality and the commonest identifiable cause of mental handicap. All primary care teams are faced with the challenges of the condition. Nothing can be done about the underlying impairment but the associated disabilities require active management if serious handicaps are to be minimized. In a condition which often requires the intervention of many specialists the role of the general practitioner is crucial — we must be aware of the common hazards of the condition if we are to prevent unnecessary morbidity. The same model of care should be applied to all mentally handicapped patients.

Incidence and aetiology

Down's syndrome is found in all races and all levels of society. About 95% of those affected have an extra chromosome 21. The incidence of Down's syndrome is strongly influenced by maternal age — for mothers under the age of 30 years the incidence is one in 2500, for those aged 29–34 years it rises to one in 1200 and in those aged 35–39 years it reaches one in 200. There is a peak incidence of one in 35 in mothers aged 39–47 years but the incidence falls for those who conceive above the age of 47 years.[1] Contrasting with this is an increased frequency of affected babies in the very youngest mothers.[2] Although maternal age is of paramount importance 20% of all Down's syndrome births are independent of maternal age and chromosomal banding shows that 30% of all meiotic errors responsible for trisomy 21 are of paternal origin.[3] Although the differences between male and female meiosis would suggest that paternal non-dysjunction is not age dependent, in reality a significant paternal age effect does occur.[4,5]

Translocations, usually with the additional chromosome attached to chromosome 14, occur in 4% of cases with Down's syndrome, with mosaicism occurring in 1% of cases.[6] In half of the translocations this is a *de novo* event in the embryo with the parents having normal karyotypes. In the other half one parent, almost always the mother, will have only 45 chromosomes with a balanced translocation, that is 45XX, −14, −21 +t (14q 21q). The theoretical risk that an affected mother will have a Down's syndrome child is one in three, but in practice the risk is one in 10. If the father carries the translocation the practical risk is only one in 20.[7]

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Mosaicism is the result of non-dysjunction in the fertilized zygote: a few cells have two cell lines, one normal and the other with 47 chromosomes. There is a marked variation in the relative proportion of each cell line within organs and tissues and if there is a relatively low number of cells with trisomy 21 in the brain then intelligence may be within the normal range. Some people with mosaicism may be difficult to identify as having Down's syndrome.

Clinical presentation and management

Occasionally, at birth there may be some uncertainty about the diagnosis of Down's syndrome and in this event the karyotype should be determined. The diagnosis is usually made by the paediatrician who would normally reveal his findings only when both parents are present. Down's syndrome is instantly recognizable to health care professionals and many of the features, for example abnormalities of the ears, saddle nose, macroglossia, strabismus and pasty dry skin represent therapeutic opportunities.

Mental impairment is virtually a constant feature among people with Down's syndrome. They score badly when subjected to formal intelligence testing and are often labelled as severely mentally handicapped. However, a low intelligence quotient is insufficient to substantiate the diagnosis of mental handicap; social functioning must also be impaired. Furthermore, when assessing social function the doctor should not only consider existing social skills but also whether the person has had appropriate opportunities for acquiring them. The majority of people with Down's syndrome will, if they are provided with good health care, emotional security and early education, fall into the mild or moderate categories of mental handicap.[8] There is considerable variation in ability and the acceptance of a low upper limit should be discouraged as this plays into the hands of those education authorities who wish to exclude children with Down's syndrome from mainstream education.

Aesthetics of Down's syndrome

'Beauty is a greater recommendation than any letter of introduction.' Aristotle

Although mental handicap is the most critical of all the disabilities in Down's syndrome it is often the facial appearance which prevents acceptance by society.[9] The plastic surgeon can help to overcome this problem. First, by reducing or removing the aesthetic and functional handicaps caused by macroglossia and secondly, by correction of the distorted facial appearance. The main stigmatizing features are a habitually open mouth, a drooping lower lip, an ugly large furrowed tongue protruding from the mouth, a flat nasal bridge which results in epicanthal folds, hypoplastic ears, a flat facial profile and a fat neck.

Olbrisch[9] advocates five procedures: partial resection of the over-sized tongue; correction of the dropping lower lip; lifting of the flat nasal bridge; correction of the hypoplastic ears; and correction of the fat neck. No plastic material is used; the flat nose is built up with homograft cartilage. All the procedures can be carried out in one operation under general anaesthesia and require only a few days in hospital.[10] Following surgery, complete closing of the mouth is possible in the majority of cases, the likelihood of respiratory tract infection is substantially reduced and articulation is usually improved. Not all children with Down's syndrome require plastic surgery but the procedures outlined are relatively risk-free and simple, so no child should be deprived of them.[10]

The problems of achieving progress in the medical management of Down's syndrome were neatly illustrated in the *Lancet* in 1983. Rozner, a plastic surgeon, presented an enthusiastic case for the benefits of surgery.[11] However, a leading article in the same issue[12] commented that the main problem of Down's syndrome is severe mental handicap, that most of those affected have an intelligence quotient of 25–50 and that surgeons can implant chins but not intelligence. These remarks were made even though Rozner had quite rightly pointed out that the procedures which he advocated were all standard; the only novelty was their use for the relief of the stigmata of Down's syndrome. Many doctors believe that because nothing can be done for the biological impairment it is not worthwhile doing anything for the associated disabilities.

Most authors emphasize the pre-school years as the most favourable time for surgical repair. Parental satisfaction is important, and 95% of parents whose children have undergone surgery would recommend the operation to other parents of children with Down's syndrome. However, the improvement in speech secondary to the correction of the macroglossia may be more immediately gratifying to the child.[13] Some parents will say that they do not wish an improved facial appearance as people will then expect their children to be more competent than they are. Perhaps the answer to this is that their children will indeed be more socially competent.

Dermatological problems

The skin of those with Down's syndrome is frequently responsible for much aesthetic handicap and functional disability. Although in infancy the skin is usually soft it soon becomes dry, thick and rough with patchy lichenification. Circumscribed sore redness of the cheeks, cutis marmorata and xerosis are frequent problems, and acne is often persistent. Recurrent furunculosis of the thighs, especially of the inner surfaces is often troublesome and cheilitis and blepharitis are sources of discomfort and unsightliness. Periocular syringoma, small dermal papules, usually flesh, yellow or mauve in colour, are frequent in adolescents. There is a high incidence of alopecia areata and vitiligo while alopecia totalis is common. These three conditions are the result of immunological deficiency in thymus dependent (T cell) function which exists in Down's syndrome. A chronic dark red, follicular dermatosis involving the presternal and interscapular skin has been found in almost half of male Down's syndrome patients (Howells G, personal observation) but the condition is rare in females.[14] Elastosis perforans serpiginosa is said to be common in Down's syndrome but in a series of 115 cases the condition was found only once (Howells G, personal observation). Hyperkeratotic patches of scaly thickened skin, greyish in colour on the limbs, are common as are keratosis palmaris and plantaris. There is also a high incidence of fungal infections of the skin.

Dry skin can be helped by the regular application of emollients and the emollient effect can also be achieved by bath oils and the regular use of soapless soaps. A dry centrally heated home environment can be improved by some form of humidification.

Sensory impairment

Impairment of hearing and/or vision can lead to an erroneous diagnosis of mental handicap even in normal children. It is not surprising then that in Down's syndrome such impairments, which occur with alarming frequency, can lead to a gross distortion in the perceived potential of an affected individual. Furthermore, unrecognized deafness may result in behavioural disorders or even apparent psychotic features, especially among those with a poor level of communication.

Hearing problems

In 1968 Fulton and Lloyd found that 58% of a group of Down's children had a significant hearing loss. Among those affected 55% had conductive loss, 23% sensorineural loss and 22% a loss of mixed type.[15] With increasing age the ratio between conductive and sensorineural loss in Down's syndrome reverses — among those aged 21 years or over 55% may have sensorineural loss. Clearly people with Down's syndrome need careful audiological supervision throughout their lives and all those involved in caring for these people must be aware of the extent of the problem and oppose the notion that a reduced hearing level may be adequate for someone with Down's syndrome. Routine testing, appropriate to the level of understanding of the individual, can be carried out in hospital audiology departments or in the case of children, at assessment centres.

Among children, the most commonly encountered problem is middle ear effusion associated with a flat tympanogram and conductive hearing loss. The tympanogram does not usually revert to normal during remissions, unlike the tympanogram of normal children. Owing to the chronic nature of the middle ear effusion, antibiotic therapy is not usually helpful. In the short term surgery may be useful but in the long term the results are discouraging — the fluid in the middle ear is so tenacious that grommets and T-tubes become blocked and their repeated removal results in an unacceptable scarring of the tympanic membrane. As the auditory canal is stenosed the surgeon often has difficulty placing the tube but even so Strome in his extensive study of the problems advocates conservative treatment only when absolute certainty exists as to the ability to follow the pathology and accurately assess its resolution.[16] In Strome's study factors relating to intelligence were explored with the conclusion that a direct relationship exists between improved hearing and improved intellect.

The indications for tonsillectomy in Down's syndrome are limited. Should the size of the tonsils become a consideration for removal, significant obstruction of the oropharynx must be present.[16] Tonsillectomy is one of the commonest procedures requested by parents and in general should be resisted. It may, however, be necessary in the management of sleep apnoea which will be discussed later.

Fortunately postaural hearing aids can be effectively worn by children with Down's syndrome. Associated abnormalities of the external ear may present difficulties in the preparation of a suitable mould, but these can usually be overcome.

Common eye problems

Virtually all adults and children with Down's syndrome suffer problems relating to the eyes but fortunately many of these are amenable to treatment. Blepharitis occurs frequently and may be the result of an abnormality of the tears.[17] Constant rubbing of the eyes often produces ectropion, entropion and trichiasis. Frequent gentle cleansing of the eyelids using warm water is helpful and acute flare-ups can be controlled by topical antibiotic eye ointments.

Most authors state that by the age of 12 or 13 years, 50% of children with Down's syndrome have cataracts but these are usually flake-like opacities which do not affect vision significantly until later life.[18] Congenital cataracts are uncommon in the general population but affect 1–5% of babies with Down's syndrome.[18] Because of their extent and density they often require early removal.

Myopia, hypermetropia and astigmatism are very common in those with Down's syndrome. Vision can usually be tested

using standard charts but when this is not possible retinoscopy using homatropine can detect myopia. The pupil in Down's syndrome appears to be sensitive to atropine, dilating quickly and remaining dilated for longer than normal. In addition, nystagmus is common and can make sight testing difficult. It often occurs only when one eye is closed so the patient should keep both eyes open during examination.

Squints, almost invariably of the convergent type, are common among those with Down's syndrome and are usually managed by correction of the refractive error. When this is not possible surgical correction is necessary. Improved vision is relatively easy to achieve but binocular function and a satisfactory cosmetic result are more difficult.

Keratoconus occurs in 5% of people with Down's syndrome.[19] The incidence is higher than in any other condition and it often appears at puberty when it may be associated with cataract and glaucoma. The acute type is painful and requires admission to hospital. The chronic form presents in a less dramatic way as increasing astigmatism.

The possibility of a visual problem should always be considered when a child or adult with Down's syndrome loses interest or becomes frustrated with daily activities which require a reasonable level of visual acuity. Ideally all babies with Down's syndrome should be seen by the ophthalmologist within the first year of life and again before they start school.

Cardiac problems

The incidence of congenital heart disease in the population as a whole is less than 1%[20] but the incidence in those with Down's syndrome is at least 50%.[21] In addition, there is a high frequency of serious abnormalities which account for 30–35% of deaths.[22] In the general population atrioventricular canal defect represents only 2% of all congenital cardiac defects but in Down's syndrome it is the commonest anomaly accounting for 60% of the cardiac findings at autopsy.[23] In people with Down's syndrome the common heart defects differ in expression and in frequency from those in the general population — in only two of a series of 300 Down's syndrome children with ventricular septal defects were the defects considered small and only one of these closed spontaneously.[24] Interestingly, pulmonary and aortic stenosis and coarctation of the aorta, which are relatively common in the general population, are infrequently met in Down's syndrome.[20]

The diagnosis of heart disease in Down's syndrome is often unnecessarily delayed. The diagnosis should be considered in the absence of a murmur as an atrioventricular canal defect may be murmur free. Often the symptoms of heart failure in Down's syndrome are erroneously attributed to developmental delay and hypotonia which is commonly found in this syndrome. It is good practice to consider the possibility of a cardiac defect in all cases of Down's syndrome; and in all cases where there is doubt a cardiological opinion should be sought.

Those caring for people with Down's syndrome should be aware of the importance of antibiotic prophylaxis for endocarditis before dental work is carried out. While recognizing that virtually any dental procedure can produce transient bacteraemia the report of a working party of the British Society for Antimicrobial Chemotherapy recommended prophylactic antibiotics only for extractions, scaling or surgery involving the gingival tissues.[25] The problem in Down's syndrome is compounded by the extent of periodontal disease, which has been reported to begin before the age of five years and to affect nearly all adults.[26] The report also draws attention to the need for prophylaxis during endoscopy or in surgery of the genitourinary or upper respiratory tract.

Respiratory problems

Children with Down's syndrome are predisposed to frequent infections of the respiratory tract, probably as a result of a poor immunological defence mechanism.[27] The lungs of those with Down's syndrome are hypoplastic, irrespective of whether heart disease is present and there is a persistence of the double capillary network in the alveolar wall which is found in the normal fetus with a diminished number of alveoli and a small alveolar surface area.[28] These anatomical changes predispose the child not only to infection but also to the development of pulmonary hypertension. Pulmonary hypertension may also result from chronic upper airway obstruction.[29]

In Down's syndrome there are two major factors which can cause obstructive sleep apnoea: first the pharynx may be crowded by lymphoid hyperplasia and the large tongue, and secondly the pharyngeal muscles are frequently hypotonic. Children who are overweight or catarrhal are particularly vulnerable. Direct effects during sleep include restlessness, unusual postures and snoring, and apnoeic pauses often result in bed wetting. Common daytime symptoms include difficulty in waking, somnolence and difficult behaviour.[30] Sleep-related upper airway obstruction is an often undetected complication of Down's syndrome and all necessary measures should be taken to overcome the obstruction before it reaches the stage of producing hypoxaemia.[31]

Alzheimer's disease

It is said that for those with Down's syndrome the reward for survival beyond the age of 40 years is presenile dementia.[32] The incidence of Alzheimer's disease is high,[33-35] but fortunately most people with Down's syndrome do not show the clinical features of Alzheimer's disease.[36] To what extent this is related to the difficulty of recognizing subtle changes in memory and performance in individuals who have few demands placed on them is difficult to know. Among 35 people with Down's syndrome over the age of 40 years only two cases of clinical Alzheimer's disease could be identified (Howells G, personal observation). They may all have had the neuropathological changes of Alzheimer's disease, but dementia is essentially a symptomatological diagnosis in which there is loss of intellectual functioning severe enough to interfere with occupational or social skills. Wisniewski and Hill have developed a useful questionnaire for assessing dementia in mentally handicapped people. It consists of a series of questions which assess items related to cognitive functioning.[37]

It is important that dementia should be considered in any middle-aged person with Down's syndrome who has developed behavioural problems, memory loss, mental inflexibility or a general slowing in activity, not only because of the possibility of Alzheimer's disease but also because of the chance of a remediable medical cause. It is often taught that delirium is common among infants and the elderly; it would be useful to add patients with Down's syndrome to the list. If a previously competent patient with Down's syndrome becomes confused a sedative is often prescribed, whereas it would be much more sensible to take the patient's temperature and carry out a proper physical examination.

Depression

Not surprisingly depression can remain undiagnosed in patients with Down's syndrome. They will have difficulty in describing the usual symptoms of depression such as lowered mood or feelings of worthlessness, and the hypochondrical delusions they describe will often sound highly improbable. Depression should always be considered when Down's syndrome patients who have

been reasonably active become withdrawn, sit alone or do not involve themselves in their usual activities. The common physical accompaniments of depression will be present, for example, poor sleep, poor appetite and loss of weight. The diagnosis is important as the condition will usually respond to conventional methods of treatment.

Bereavement

Bereavement poses many problems for patients with Down's syndrome. Many live with elderly parents and the degree of attachment and dependence may be strong. McLoughlin argues that the mentally handicapped, because of their reduced social horizons restricting the development of peer group relationships, may have a great investment in a few highly significant relationships; the breaking of these close bonds may be catastrophic.[38] Often, people with Down's syndrome are unable to understand death or to negotiate the normal mourning processes. Bicknell in her sensitive account of the psychopathology of handicap describes how a handicapped person is often sent on a fortnight's holiday when there is a death in the family while the rest of the family grieve the loss. The handicapped person is assumed not to notice the loss[39] but exclusion from the grieving process often results in overlooked depression and behavioural difficulties.

As people with Down's syndrome are now living longer this problem will increase. When the carers are aged or in poor health the handicapped person should be introduced into alternative accommodation, perhaps on a regular short term basis so that new relationships can develop. Not unexpectedly carers may be reluctant to agree to these changes as so often they are emotionally dependent on the attachment themselves. It is a situation that requires sensitivity and understanding on the part of the general practitioner.

Epilepsy

Epilepsy is less common among people with Down's syndrome than in the mentally handicapped population as a whole.[40] The frequency of fits is less than 2% in Down's syndrome patients under the age of 20 years but it rises to 12% in those aged over 55 years suggesting neurological degeneration.[41] There are special problems in treating epilepsy in patients with Down's syndrome. For example, the clinical signs of toxicity from anticonvulsant drugs are difficult to elicit and so the monitoring of blood levels is particularly important.[42] Antiepileptic drugs can cause serious degradation of cognitive function leading to a syndrome resembling dementia — phenytoin is more likely to do this than other antiepileptic drugs causing 'phenytoin encephalopathy'.[43] Of the antiepileptic drugs phenytoin has also been shown to have the greatest effect on attention and memory with sodium valproate and carbamazepine having the least effect.[44] In contrast with normal adults who will readily complain when drugs are causing unpleasant symptoms, people with Down's syndrome are often unable to complain and are obliged to continue with medication.

Thyroid disease

Several studies have shown that there is an increased prevalence of hypothyroidism among people with Down's syndrome compared with the general population.[45] In a series of 52 adults with Down's syndrome attending adult training centres seven were found to have biochemical evidence of hypothyroidism (Howells G, personal observation). Interestingly 18 of the total were found to have a T4 level in the lower normal range. In only one case was a diagnosis of hypothyroidism made on clinical grounds. Part of the difficulty is that in general those with Down's syndrome are slower and less active than normal people, have thickening of the skin, have difficulties with articulation, which masks any hoarseness, and are less alert. Murdoch and colleagues initiated a study on the basis of an observation that the electrocardiograms in adults with Down's syndrome commonly show abnormalities which are consistent with hypothyroidism.[46] In a study of 82 adults with Down's syndrome they found that 40% had abnormalities of one or more thyroid functions suggesting underactivity: hypothyroidism had been suspected in only one case. In contrast, 50% of a group of 55 adults with Down's syndrome resident in a hospital for the mentally handicapped were thought to have clinical features suggesting hypothyroidism.[47] Clearly, people with Down's syndrome must be screened for hypothyroidism at least every five years and thyroid antibodies should also be checked. A positive antibody test indicates that hypothyroidism will almost certainly develop.[48]

Hyperthyroidism is rare in patients with Down's syndrome but is important because it can present as a behavioural problem.[49] With the increasing life span of people with Down's syndrome general practitioners must become more aware of the possibility of an organic brain disorder such as hyperthyroidism and the need for specific treatment.

Joint and muscle problems

Atlantoaxial instability was first described in 1830 by Bell.[50] Recently because of the greater involvement of people with Down's syndrome in sporting activities, potential dangers of the condition have been highlighted.[51] The abnormality consists of a misalignment of the first two cervical vertebrae and during hyperextension or flexion of the neck this presents a neurological hazard. The condition affects between 12 and 22% of all people with Down's syndrome. It might appear reasonable to screen for atlantoaxial instability when patients are five or six years old but the clinical significance of this radiographic sign is not fully understood and thus it is not justifiable to ban affected patients from activities which are socially rewarding and physically beneficial. Perhaps of greater clinical importance is the high incidence of degenerative cervical arthritis in young people with Down's syndrome, often associated with the clinical signs of myelopathy. Because of the inherent problems of communication, people with Down's syndrome are in general unable to describe the resulting symptoms.[52]

Obesity

Obesity is common in patients with Down's syndrome and the problem appears to be more marked from early childhood to adolescence. Not only is the motor development of the child hampered but also the social development as a result of restricted recreational activities with other children. The parents of infants with Down's syndrome should be warned of the potential problems ahead so that they can form appropriate eating patterns. Established obesity in Down's syndrome is difficult to manage and requires persistence over a long period.

Nutritional supplements

General practitioners will frequently be asked about the value of mineral and vitamin supplements for patients with Down's syndrome. A well organized clinical trial was carried out in Chicago to determine whether supplements improve the level of intellectual functioning. The results demonstrated that the use of a combination of nutritional supplements in school-aged children with Down's syndrome did not lead to improvements in intellectual test performance.[53,54] Although there is no specific evidence that megavitamin therapy (doses vastly

exceeding the amount recommended for nutritional balance) has a place in the management of Down's syndrome there is a great deal of anecdotal evidence that the general health of this group is often dependent on vitamin and mineral supplementation. It would seem reasonable to provide vitamins A, B, C and D together with a zinc supplement.

Screening for Down's syndrome

The present policy on antenatal screening for Down's syndrome is to offer amniocentesis to all women who will be aged 36 years or over at their expected date of delivery. Screening is also offered to couples who have already had a child with a chromosomal abnormality or who are known to be carrying a balanced translocation. Unfortunately, screening based on age alone is unsatisfactory and has had no significant influence on the incidence of Down's syndrome at birth. Amniocentesis in the 5% of expectant mothers who are over 36 years of age will only detect 30% of all Down's syndrome pregnancies. Moreover in practice less than 15% of affected pregnancies are detected because less than half of the older women take up amniocentesis.[55] The general practitioner has a role to play in trying to improve this figure. Sometimes a history of threatened abortion is given as a reason for omitting amniocentesis despite the fact that bleeding in the first trimester occurs in 26% of women carrying a Down's fetus compared with 1% of mothers carrying a normal fetus.[56]

Pregnancies resulting in babies with Down's syndrome are associated with reduced maternal alpha-fetoprotein concentration.[57] More recently a method of detecting Down's syndrome, measuring human chorionic gonadotrophin, unconjugated oestriol and alpha-fetoprotein in the maternal serum at 16 weeks has been found to be reliable.[58] Taking into consideration maternal age, this method of screening could detect over 60% of affected pregnancies. Such a programme could reduce the number of children born with Down's syndrome in the United Kingdom from about 900 a year to about 350.[58] It is likely that this cost effective method of screening will gradually become standard. Some patients who are recognized to be at high risk may still prefer to have chorionic villi biopsy or amniocentesis early in pregnancy because of the more certain diagnosis which these techniques offer.[59,60] The difficulty for general practitioners is to balance the wishes of an individual woman and her personal risk against the benefits of population screening programmes with high rates of detection. It has been suggested that the new screening strategy should apply for all women aged under 38 years and efforts made to provide chorionic villi biopsy for women above that age.[61] Naturally the decision to have amniocentesis or chorionic villi biopsy has to be made by the couple and the responsibility of the general practitioner is to provide them with up to date information free from any personal prejudice.

It is essential to know the chromosomal type in every child with Down's syndrome not only to assess the risks to siblings but also the risk to other close relatives. When the chromosomal type of someone with Down's syndrome is unknown then a patient who is a relative of that person must have his or her chromosomes checked to exclude a translocation carrier state.

Prognosis for Down's syndrome

The social prognosis for Down's syndrome has improved dramatically over the past few years. Until recently society considered children with Down's syndrome to be ineducable and incapable of a meaningful life but now they can learn to read and to care for themselves and can acquire skills in some trades; many can live semi-independently with minimal supervision. However, the medical prognosis is not as optimistic. The high incidence of congenital heart disease and immunodeficiency result in a high mortality in infancy and childhood and in those aged 40 years and over — Øster found that the mortality rate for respiratory disease was very high, especially for the age group five to 14 years.[62] In those aged over 40 years there is a high incidence of neurological disorders and the mortality rate for premature ageing and strokes is five times higher than in the general population. Although there is evidence that Down's syndrome confers a greater than 10 fold increased risk of leukaemias in children, it is not known if adults have comparable increased risks.[63] One prospective study did, however, show that the mortality rate from miscellaneous cancers was five times higher among men with Down's syndrome than among men in the general population.[64]

Care in the community

This review has suggested ways in which the general practitioner can respond to some of the medical challenges of Down's syndrome. We must ensure that the medical needs of Down's syndrome patients are not overlooked and that referrals to the specialist services are appropriate. At the same time the adoption of the clinical perspective must not isolate people with Down's syndrome from other professional and voluntary agencies. Although the management of disability and the prevention of handicap remains the primary role of the general practitioner it is essential that a therapeutic relationship is established with the families of Down's syndrome patients. People with Down's syndrome and their families experience the same life events as everyone else and often these are periods of intense emotional turmoil. The birth of a sibling, starting school, the onset of puberty and leaving school are times when regression, frustration or maladaptive behaviour may appear. The out-of-home placement is probably the most important psychosocial event and those with Down's syndrome usually have little say in where they are placed. Whereas the non-handicapped adult may experience a new freedom from parental demands, the handicapped person leaving home may, paradoxically, experience new and greater restrictions. Thus, the grief at leaving home may be greater than anticipated because it is not tempered by any gains.[65] Now that community based services are established with a greater variety of out-of-home placements, separation from the family, can, if carefully planned, become a positive growth experience. Residential care should be accepted as a normal life event and not seen as rejection by the family. The general practitioner, if he is to participate in these processes must see himself as part of a larger network of services providing care for people with Down's syndrome and their families. Studies of the clinical aspects of Down's syndrome reveal large discrepancies between those living in institutions and in the community. For example, studies show that institutionalized people with Down's syndrome have a much higher incidence of hepatitis carrier status than other mentally handicapped people.[66,67] However, a study carried out on a group of people with Down's syndrome living in the community showed that they do not have a higher incidence of positive hepatitis B antigen than the general population.[68] Thus, patients with Down's syndrome who have never been institutionalized should be treated no differently from any other person in the general population who is not suspected of being a transmitter of infectious disease.

In this paper it has not been possible to review the hundreds of recent articles on Down's syndrome and so attention has been focussed on those topics of greatest practical importance. We have created many of the images of Down's syndrome patients ourselves; we must now reverse the negative images of this intriguing condition.

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