

# INFANCY TO ADULTHOOD WITH DOWN'S SYNDROME

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## INTRODUCTION

Most of our knowledge of Down's Syndrome – single palmar creases, epicanthic folds, flat occiput, duodenal atresia, birth prevalence of 1.6 per thousand – is of little help to ourselves or our patients. "Can I go horse riding?" "Will my child go to the local primary school?" "Who will look after my son if I die in my seventies?" These and many others are the questions which individuals and families ask.

## KNOWLEDGE, ATTITUDE AND PRACTICE

The quality of life is improving and life expectancy for people with Down's Syndrome and their families is increasing. This is partly because of increased knowledge but is more the result of changing attitudes and practice of professionals and society. For example, families have taught us to show respect for people by talking of a child with Down's Syndrome, rather than "mongol", "the Down's" or "the Down's child". The impact on a family of having a child with Down's Syndrome is so profound that I recommend that we speak of a *family* with Down's Syndrome just as we speak of a person with Down's Syndrome.

Down's Syndrome is identified by a relatively small number of items. Each individual has many thousands of unique features and the differences between people with Down's Syndrome massively exceed the similarities. It is helpful to remind families that their child has a unique combination of genes inherited from both parents and is therefore like no other child.

Studies have demonstrated that doctors looking after children with identical cardiac lesions, or with leukaemia, are less encouraging to families with Down's Syndrome than to those with an otherwise normal child, and that medical or surgical treatment is different. In a similar way the birth of a child with Down's Syndrome is frequently perceived by those involved with the pregnancy as a failure of antenatal care. Our assumptions of normality and the prejudices surrounding handicap should be challenged. Parents experience and show a normal grief reaction for the loss of the normal foetus/child. The denial, anger and blame are normal and transient. Like nearly all children these newborns become a special part of their family. It is after their grief that we must ask people if the antenatal service has failed them.

The details of how parents are told that their child has Down's Syndrome are always indelibly etched into their memories in vivid detail. This major life event has often been incorrectly managed and parents often remain understandably angry for the remainder of their lives.

The 1981 Education Act was a significant step and

## BREAKING THE NEWS

- 1 News to be given by experienced and knowledgeable consultant paediatrician, accompanied by one other (social worker, midwife, health visitor). It often helps to start positively with "I have some news for you that you were not expecting."
- 2 Tell the parents as soon as possible
- 3 Both parents to be seen together, or mother plus supporter (eg child's grandmother)
- 4 To be in private, without interruption – schedule one hour
- 5 To have baby present, referred to by name. Convey warmth, understanding and respect
- 6 Give truthful and accurate information
- 7 Help the parents to pass the news to family and friends
- 8 Have follow-up meetings; the first usually within 24 hours
- 9 Provide a written summary of the discussion, and appropriate support materials, contacts etc.

Figure 1

reflected changing attitudes: prior to this, schools for children with severe learning difficulties were part of the health service. The new act recognised that the right to education is independent of health, impairment and handicap. Each child should be enabled to reach his maximum potential by providing the most appropriate education. Every child requiring special education has his needs assessed and the necessary input provided either within mainstream education or at a special school.

## DEVELOPMENT, INTELLIGENCE AND EDUCATION

In infancy and the preschool years, children with Down's Syndrome demonstrate developmental delay, and usually social development is less affected than other skills. All these children have learning difficulties ranging from mild to severe, but parents can be assured that their child will become continent, mobile, able to communicate and enjoy life. The strongest determinant of a child's level of attainment is his parents' level of education. Children may not reach their full potential if professionals fail to support families or to provide appropriate education.

Caution must be exercised because excessive input (as in some intensive intervention programmes) does not increase the level of an individual's potential. There is no evidence that nutritional treatments alter this development potential.



Most of these children should be placed in mainstream infant school with the necessary support prescribed in the Statement of Special Educational Need. Some pupils with Down's Syndrome will remain within mainstream education through secondary school and occasionally into further education. As they get older, however, pupils will usually require the resources of a special school. The most able adults with Down's Syndrome will attain a reading age of 10-12 years and a comprehension age of about eight years. Only 40% will achieve reading as a skill, and few will get beyond the levels of addition and subtraction.

## MEDICAL PROBLEMS

Down's Syndrome is the result of extra chromosomal material present in 50-100 genes on the extra chromosome 21, whether from trisomy or translocation. In the foetus this extra gene dosage increases the potential for disorder during organogenesis.

Thereafter individual genes in overdosage seem to lead to an increased rate of ageing and to impaired immunity, which underlie many of the problems experienced from infancy onwards.

### Cardiorespiratory system

Forty to fifty percent of children with Down's Syndrome are born with heart disease. This is sixty times the normal prevalence. The most common abnormality is atrio-ventricular canal defect. Congenital heart disease accounts for 30-35% of deaths in Down's Syndrome and the mortality is highest in the first two years of life. Heart disease, abnormal morphology of the respiratory system and impaired immunity cause an increased risk of chest infections and susceptibility to cor pulmonale. Congenital heart disease is not always clinically apparent at birth and all neonates require echocardiography.

### Vision

Forget epicanthic folds and Brushfield's spots: although well known, they present no problems. More importantly, many individuals require spectacles, often commencing in infancy: the majority of children develop a degree of hypermetropia, up to 20% may show myopia and a small percentage astigmatism. Frequently non-patent lacrimal ducts cause conjunctivitis, and tear spillage which exacerbates the facial skin problems. The duct is easily made patent. Up to 50% of individuals have cataracts. These are usually on the periphery of the lens and fortunately rarely affect vision. Visual assessment is necessary in early infancy and annually thereafter.



### Hearing

There is significant hearing impairment in approximately 50% of children with Down's Syndrome. The commonest problem is conductive middle ear disease caused by a combination of impaired immunity and the abnormal anatomy of the pharynx and eustachian tubes. The altered anatomy can also result in upper airway obstruction and sleep apnoea. Slightly less than half of the hearing impairment is a sensori-neural problem and all newborn children with Down's Syndrome must have careful assessment.

### Growth and endocrinology

Growth retardation is a feature of Down's Syndrome, possibly caused by organ resistance to hormones and deficiency in insulin-like growth factors. Growth charts have been produced which demonstrate the normal growth pattern of children with the condition. There is an excess of thyroid disease and diabetes compared with the normal population. The subtly altered immune function in Down's Syndrome has not been identified but is likely to be instrumental in these endocrine diseases. The natural history of thyroid antibodies and thyroid status in Down's Syndrome is not yet fully worked out. Transient raised levels of thyroid stimulating hormone with normal levels of thyroxine usually return to normal but occasionally progress to frank hypothyroidism. The sensible pragmatic approach is to be alert to endocrine disease and to measure thyroid function at intervals of three years from the age of ten.

### Neurology

Neurological problems are commoner in Down's Syndrome. Epilepsy, the commonest neurological disease in childhood, has a prevalence of between 1% and 5%, compared to 0.5% in the general childhood population.

The Department of Health produced guidelines restricting the range of activities for people with Down's Syndrome because of possible atlanto-axial dislocation of the cervical vertebrae. Approximately twenty cases of spinal cord damage are reported in the world literature. It is **NOT** appropriate to curtail normal activities such as riding, football and swimming. Common sense should be used and families given accurate information when their child wants to pursue active sports such as trampolining or diving. Anaesthetists and others must be very cautious when manoeuvring the head and neck of an unconscious or paralysed person with Down's Syndrome (for example at intubation) so as to avoid damaging the spinal cord.

The gene locus for the pre-senile dementia of Alzheimer is located on chromosome 21. The onset of manifestations occurs earlier in Down's Syndrome, and increasingly so after the age of forty. At least one third of adults with Down's





Syndrome will develop Alzheimer's and perhaps many more than this. The increased care burden often falls on parents who are themselves becoming older and frail.

### Gastrointestinal problems

The management of constipation, which occurs frequently, is the same as for anyone, including encouraging plenty of fluids, fruit, vegetables and cereals, and the use of simple laxatives.

### Dermatology

Skin problems are common. The skin is often dry with seborrhoeic dermatitis of the scalp and a wind-chapped face often exacerbated by tears. Up to 75% of people with Down's Syndrome have hyperkeratotic areas particularly on the upper limbs. The use of emollient on the skin and emollient bath additives is helpful.

### Haematology

The incidence of leukaemia is higher as are the morbidity and mortality rates. Patients are more susceptible to infection and are more sensitive to drug toxicity. There is also concern that these patients are not being treated in the same way as children without Down's Syndrome.

### Life Expectancy

Life expectancy is reduced, mainly because of the incidence of severe congenital heart disease. The majority of babies born with Down's Syndrome will live to their fourth decade. A study in the UK fifteen years ago showed the average life expectancy to be into the sixth decade: therefore more years of care occur in adult life than in childhood.

Life expectancy	Survival			
	5 years	10 years	20 years	30 years
with congenital heart disease	62%	57%	53%	50%
without congenital heart disease	87%	85%	82%	79%

Table 1 - Life Expectancy (from a recent Canadian study of 1,341 people with Down's Syndrome)

### Behaviour

The image of people with Down's Syndrome being loving, happy and music-loving is a stereotype and is an insulting way to think of any person.

Behavioural problems are more common and their management is particularly difficult since they coincide with learning difficulties. Families need support and guidance, and sometimes specialist input.

### Adolescence and adulthood

People with Down's Syndrome grow, develop and progress through the stages of life just as everyone does. Adolescence is a time of increasing independence when self-esteem and confidence are in a state of great flux. This is more marked for people with mental handicap, including Down's Syndrome. Contact with other adolescents and others in society leads to the realisation of difference and perhaps a sense of inferiority. Combined with the withdrawal of regular education and child health services at around age 18, this can lead to insecurity and isolation.

Sexual awareness is delayed in adolescents with Down's Syndrome and may never appear. Relationships with the opposite sex are common. There is less desire for sexual intercourse than in most people and relationships are usually platonic. Sexual activity is usually limited to masturbation. Some adults with Down's Syndrome do have a need for a sexual relationship and a small number marry. Fertility is markedly reduced but both men and women become parents. Families will need increasing advice on helping their adolescents and young adults through these important stages.

### SUMMARY

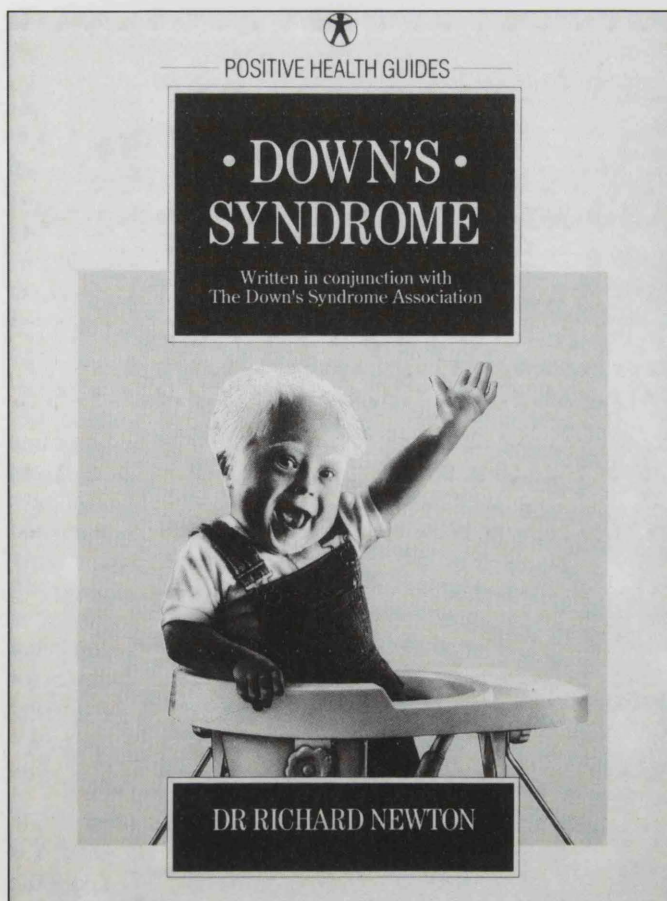
Today people and families with Down's Syndrome have an improved life compared to their predecessors 20 years ago. They are very much part of the family and society in which they live. The recent decades have brought greater acknowledgement of the need for self-determination in people with mental handicap. Much of this advance has been led by families with Down's Syndrome. Doctors and other professionals can improve their performance and image by being more positive about impairment and handicap such as those in Down's Syndrome.

#### Protocol for yearly medical review of children with Down's Syndrome (may be modified for use with adults)

- 1 Review parental worries. Include enquiries about sleep, behaviour, constipation
- 2 General medical and growth assessment (use Down's Syndrome chart)
- 3 Neurological enquiry and examination, particularly lower limb neurology and gait
- 4 Thyroid function: enquire about symptoms. From age of ten years test at three-yearly intervals
- 5 Annual hearing assessment
- 6 Annual orthoptic/vision testing
- 7 Enquire about education, finances, support







## **FURTHER INFORMATION FOR PARENTS, FRIENDS AND PROFESSIONALS**

The Down's Syndrome Association 153-15 Mitcham Road, London SW17 9PG 0181 682 4001

Lancashire branch of The Down's Syndrome Association contact Julie Maxfield 01254 849463



Advisory Centre for Education (ACE) 1 Aberdeen Studios, 22-24 Highbury Grove, London N5 0171 354 8321

Newell P ACE Special Education Handbook College Hill Press, London

Cunningham CC, Sloper P Helping your handicapped baby Souvenir Press, London 1978

Cunningham CC Down's Syndrome: An introduction for parents Souvenir Press, London 1990

Any of the Lancaster consultant paediatricians, particularly Dr Sue Brown and the multiprofessional team based at Longlands Child Development Centre. 01524

## **LITERATURE**

1 Newton RW, Newton J The management of Down's Syndrome Recent Advances in Paediatrics Number 10, David TJ (ed) Churchill Livingstone 1992

2 Lane D, Stratford B (eds) Current approaches to Down's Syndrome. Holt, Reinhert & Winston, Eastbourne

3 Newton R Down's Syndrome Optima 1992