A survey of Down's syndrome under general practitioner care in Scotland

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SUMMARY. A survey of all general practitioners in Scotland has revealed details of 1,251 individuals with Down's syndrome being cared for by their family doctor; 1,150 were living at home.

Eighty-two per cent of doctors responded to the questionnaire. High prevalence rates have been noted in the Western Isles and in Shetland. The survey confirms reports of a lowering of maternal and paternal age in Down's syndrome; 72 per cent of the mothers and 56 per cent of the fathers of Down's syndrome children born since 1971 were under 35 years at the time of birth. The survey also revealed an increase in the number of Down's people living with either one parent or relatives with increasing age. It is hoped that further research will be carried out into the problems facing Down's individuals and their families through the medium of general practice.

Introduction

THE recent public debate on the fate of a new-born child with Down's syndrome has underlined the need for information about current expectations of quality of life for individuals with Down's syndrome in the United Kingdom. The image of the Down's syndrome individual in the mind of the public is of a happy-go-lucky defective with musical tastes who is a burden to elderly parents and who is more often than not permanently institutionalized. To a certain extent the information given to parents by general practitioners follows this public view and no clear guide seems to be given as to what parents might reasonably expect of a new-born infant with Down's syndrome. Down's syndrome is a comparatively rare condition with a reported birth prevalence of between 10 and 14 per 10,000 (Smith and Berg, 1976). The purpose of this paper is to describe the findings of a survey carried out in general practice in Scotland in 1981.

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Method

A letter was sent to all 1,138 practices in Scotland asking for information about patients with Down's syndrome registered with the general practitioner. The information sought was in three main areas:

1. On the Down's individual
a) Sex and date of birth.
b) Normal place of residence.
c) Whether the diagnosis had been confirmed by chromosome analysis.
2. On the Down's family
a) Year of birth of mother and father.
b) Composition of families.
c) Parents' occupation.
d) Number of older and younger siblings.
3. On the practice
a) Total practice list size.
b) Health Board area.

Results

Replies were received from 933 practices (82 per cent). There was considerable regional variation in the response rate, ranging from 100 per cent in Shetland and more than 90 per cent in Forth Valley, Grampian, Orkney and Tayside to 66 per cent in Lanarkshire. Forty per cent of practices reported no cases of Down's syndrome.

Down's individuals

Numbers, age and sex

The age and sex distribution of reported cases under the care of their general practitioner can be seen in Table 1. Only those children aged one year or more on 1 January 1981 were included in the statistics recorded here. Figure 1 shows that the age and sex distribution was biased

Table 1. Age and sex distribution of Down's syndrome under general practitioner care.

	Age at 1 January 1981 (years)							
	1–4	5–9	10–14	15–24	25–34	35–44	45+	Total
Male								
(per cent in each group)	96 *(13.9)*	95 *(13.7)*	101 *(14.6)*	152 *(21.9)*	137 *(19.8)*	72 *(10.4)*	40 *(5.8)*	693 *(100)*
Female								
(per cent in each group)	64 *(11.5)*	81 *(14.5)*	71 *(12.7)*	142 *(25.4)*	108 *(19.4)*	63 *(11.3)*	29 *(5.2)*	558 *(100)*
All								
(per cent in each group)	160 *(11.9)*	176 *(14.0)*	172 *(13.7)*	294 *(23.5)*	245 *(19.6)*	135 *(10.8)*	69 *(5.5)*	1,251 *(100)*

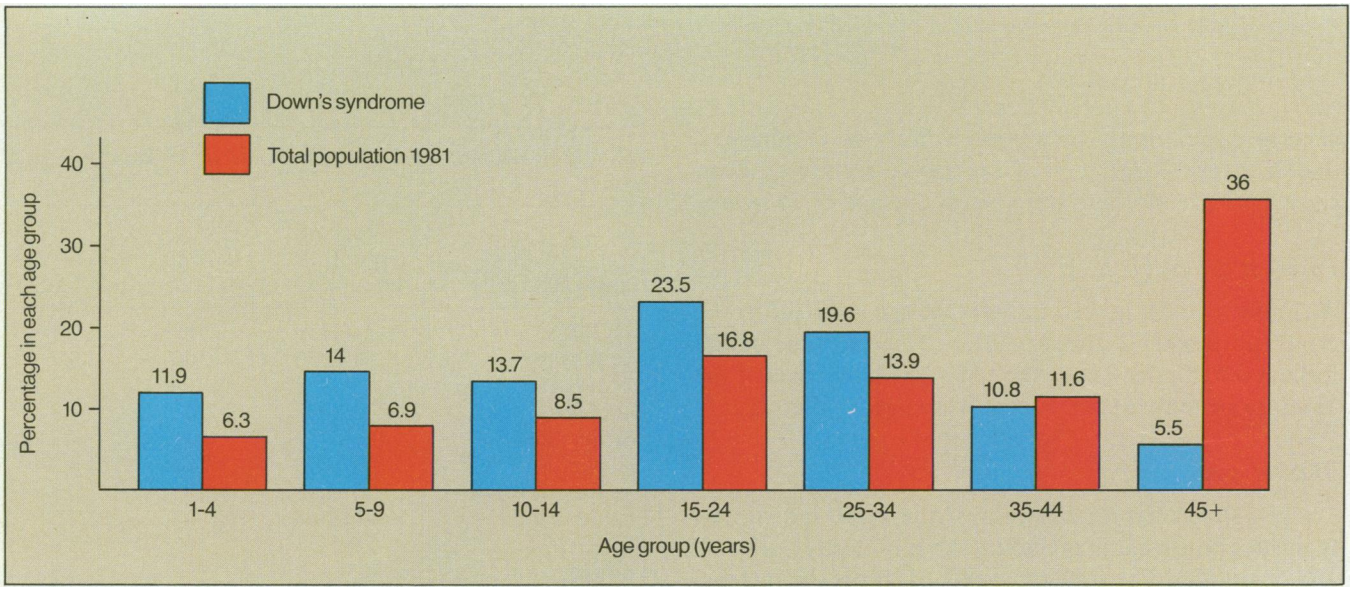

Figure 1. *Distribution of reported Down's syndrome and of the total population, by age group.*

towards the younger age groups and that only 5.5 per cent were aged 45 years or over, compared with 36 per cent in the general population.

Prevalence of Down's individuals in age and sex groups is seen in Table 2. The population denominators in each group have been estimated at 82 per cent (the overall response rate) of the projected populations for 1981 produced by the Registrar General for Scotland. The prevalence rate drops generally with age, and the male prevalence rate is higher than the female rate in all age groupings.

The prevalence of Down's individuals grouped by Health Board area is seen in Table 3. Here the practice population is used as the denominator and only Down's patients from those practices giving a population are included. A high prevalence rate is seen in the Western Isles, Shetland, Borders and Orkney. However, the number of Down's individuals in these areas is very small.

No significant increase was seen in the frequency of births in any particular month.

Normal place of residence

As might be expected with a group who are by nature community based, the vast majority (92.5 per cent) were living at home with one or both parents or relatives; the remainder were in hostels, residential schools or in hospital, but under the care of the general practitioner.

Chromosome analysis

A total of 415 individuals had had chromosome analysis (30.7 per cent). No attempt was made to identify the type of abnormality.

Down's families

Maternal age at birth

When the overall results of maternal age at birth of the Down's individuals were analysed, there was a preponderance of older mothers. However, Figure 2 demonstrates that in those born after 1 January 1971, there is a marked difference in maternal age in that there are more younger mothers. Figure 3 compares maternal age in Down's syndrome for the decade 1971-1980 with the 1975 figures for maternal age of legitimate live births in Scotland and demonstrates that there is still a preponderance of births of Down's syndrome to mothers aged over 35 (34.4 per cent as opposed to 6 per cent in the

Table 2. Estimated prevalence of Down's syndrome under general practitioner care by age and sex.

	Age (years)							
	1–4	5–9	10–14	15–24	25–34	35–44	45+	Total
Male								
Number of Down's syndrome	96	95	101	152	137	72	40	693
Estimated population	103,900	149,300	183,500	362,800	294,600	242,500	665,000	2,001,600
Prevalence/10,000	9.2	6.4	5.5	4.2	4.7	3.0	0.6	3.5
Female								
Number of Down's syndrome	64	81	71	142	108	63	29	558
Estimated population	98,700	143,100	174,200	348,200	291,800	248,900	862,300	2,167,200
Prevalence/10,000	6.5	5.7	4.1	4.1	3.7	2.5	0.3	2.8

Estimation is 82 per cent (response rate) of projected populations for 1981 (see text).

general population). However, it must be emphasized that in over 70 per cent of the Down's children born since 1971 the mothers were below the age of 35 years.

Paternal age at birth

A similar pattern is seen in paternal age at birth. In the group born after 1 January 1971, 57.6 per cent of the fathers were under 35 at the child's birth, whereas of the older group only 36.5 per cent were aged under 35.

Marital environment

Table 4 shows the distribution of the 1,150 individuals living at home (that is excluding those in hostels, schools and hospitals). The striking aspect of this is the increasing number of subjects living with just one parent or with relatives as age increases: 288 were living with one parent, and in 247 cases (86 per cent) that parent was the mother. In 184 cases the father was dead and in 63 the parents were separated. Of the 41 cases where the father was the one parent, the mother was dead in 38.

Parents' occupation/social class

Social class was ascertained by occupation of the father, where present. No differences were seen in the proportions of Down's individuals in the various social classes and reported national statistics (Annual Report of Registrar General for Scotland, 1975).

Number of siblings

Of all Down's individuals, 18.1 per cent had no brothers or sisters. The 1,018 Down's individuals whose siblings were recorded had a total of 2,315 siblings—making an average family size of 2.3; 72.9 per cent of all Down's individuals had no younger siblings.

Table 3. Prevalence of Down's individuals by Health Board areas.

Health Board	Reported practice population	Down's patients	Prevalence per 10,000
Argyll & Clyde	256,839	88	3.4
Ayr & Arran	312,276	100	3.2
Borders	94,206	54	5.7
Dumfries	123,250	53	4.3
Forth Valley	256,246	107	4.2
Grampian	371,840	111	3.0
Greater Glasgow	591,658	165	2.8
Highland	160,563	66	4.1
Lanarkshire	298,585	107	3.6
Lothian	434,131	151	3.5
Orkney	17,445	9	5.1
Shetland	19,977	15	7.5
Western Isles	23,498	24	10.2
Fife	203,595	61	3.0
Tayside	251,741	102	4.1
Scotland	3,415,850	1,213	3.6

Discussion

This survey of individuals with Down's syndrome and their families under the care of the general practitioner demonstrates that the general practitioner can be an important source of epidemiological information on a rare condition, so rare that each general practitioner will have a Down's baby born in his practice only every 38 years of his professional life. The purpose of this report is simply to record these facts and to discuss them in the light of what is known about Down's syndrome.

Males outnumber females in all age groups and the male:female ratio is 1.24:1. This preponderance of males was previously noted in a study in North East Scotland (Speed *et al.*, 1976) which quoted other such reports. The excess of males is thought to be due to an excess mortality of females in infancy from cardiac disease (Fabia and Drolette, 1970), but this has been disputed (Øster *et al.*, 1975). Interesting comparisons can be drawn between the 1981 figures for Grampian, Orkney and Shetland from this study and the study by Ross and colleagues (1967). The prevalence rate for all Down's individuals living outside institutions in 1967 was 4.2/10,000, compared with 3.3/10,000 in 1981 (the

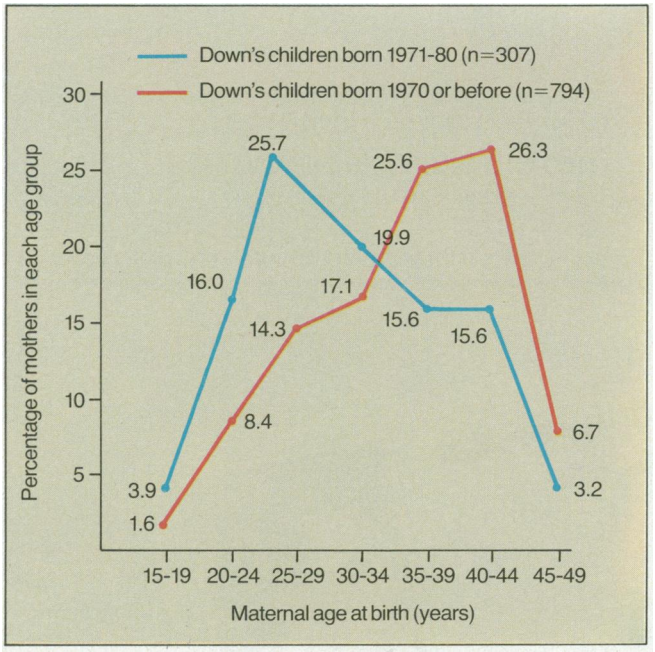

Figure 2. *Maternal age at birth of Down's children born before and after 1970 (per cent of each age and sex group).*

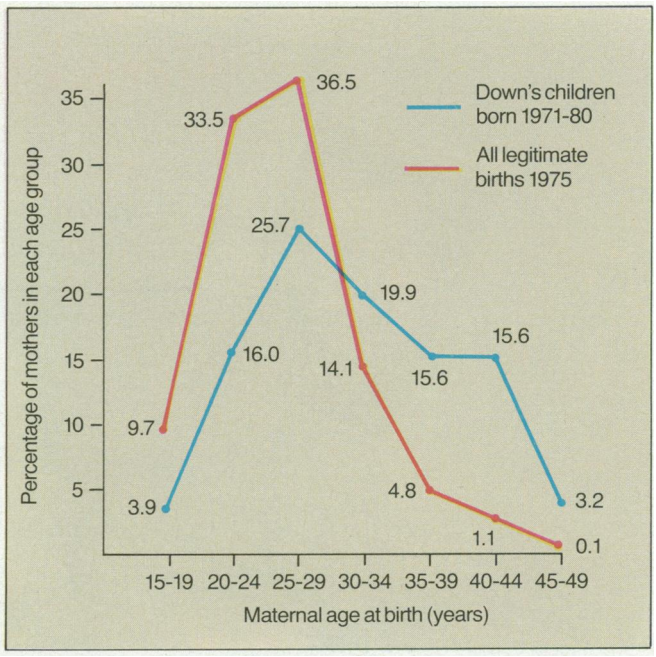

Figure 3. *Comparison of maternal age at birth of Down's syndrome children born 1971-80 and all legitimate births in 1975.*

1967 prevalence rate was corrected to include those living at home).

The interesting regional variations in prevalence referred to above are in the Western Isles and Shetland. The high prevalence in Shetland has been reported previously (Ross *et al.*, 1967), but no previous report has been made of a high prevalence in the Western Isles. Of the 41 individuals living in the Island areas, 36 are living at home (88 per cent) and 24 are aged 25 years or over. This apparent high prevalence may, therefore, simply reflect island attitudes to sending Down's relatives to remote institutions. The capacity of such remote island communities to cope with such a high prevalence rate is encouraging for those who maintain that other communities might have more potential in caring for the mentally handicapped. Of the 54 individuals in the Borders, 18 are in hostels or residential homes, and it would appear that the 'high prevalence' simply reflects the fact that here institutionalized patients are under the care of the general practitioner.

The survey confirms the findings from Manitoba (Uchida, 1970), British Columbia (Lowry *et al.*, 1976), Japan (Kuroki *et al.*, 1977), Copenhagen (Mikkelsen *et al.*, 1976), Sweden (Lindsjo, 1974), Belgium (Koulischer and Gillero, 1980), Massachusetts (Holmes, 1978) and South Australia (Sutherland *et al.*, 1979) that Down's children are now being born mainly to younger mothers and fathers. However, recent evidence from Manitoba (Evans *et al.*, 1978) and New York State (Hook and Cross, 1981) points to an increase in births to the 35–39 age group, although the proportion of births to the 35 and over group remained the same. The age of the father is equally important, since it is now known that the father is the source of the extra chromosome in 20–25 per cent of cases (Magenis *et al.*, 1977) and statistical evidence from Norway (Erickson and Bjerkedal, 1981) has shown a significant paternal age effect. In those born between 1970 and 1979 in this study, 70 per cent of the mothers and 58 per cent of the fathers were under the age of 35, whereas in those born before 1955, only 37 per cent of the mothers and 40 per cent of the fathers were under 35 at the birth of the child.

This established decrease in parental age in Down's syndrome, which apparently predated the wide application of amniocentesis, produces a dilemma for those involved in the economics of the provision of amniocen-

Table 4. Percentage of Down's individuals living at home by age group*.

Age group	Number	With both parents. Number (per cent)	With one parent. Number (per cent)	With relatives. Number (per cent)
All	1,150	789 *(68.6)*	288 *(25.0)*	73 *(6.4)*
0–4	154	144 *(93.5)*	7 *(4.5)*	1 *(2.0)*
5–9	169	147 *(87.0)*	22 *(13.0)*	0 *(0.0)*
10–14	162	135 *(83.3)*	24 *(14.8)*	3 *(1.9)*
15–24	268	205 *(76.5)*	58 *(21.6)*	5 *(1.9)*
25–34	225	114 *(50.7)*	97 *(43.1)*	14 *(6.2)*
35–44	116	37 *(31.9)*	63 *(54.3)*	16 *(14.8)*
45+	56	5 *(8.9)*	17 *(30.3)*	34 *(60.8)*

*Individuals under the care of their general practitioner but living in residential schools, hostels or in hospital have been excluded.

tesis. The present position of offering amniocentesis only to pregnant women of 35 years or over is based on cost-benefit analysis studies (Hagard and Carter, 1976). There were, however, only 4,055 births to all mothers of 35 years and over in Scotland in 1975, with an expected yield of 30 per cent of the Down's infants. However attractive this small group might appear to the geneticists and those paying for genetic services, even given 100 per cent success, the strategy is going to identify only 30 per cent of cases. A search for another factor apart from maternal age in order to select a diagnostically fruitful, and therefore an economically viable, group is, therefore, urgently required. One wonders, for example, if paternal age is at present used as a criterion for offering amniocentesis.

It must, however, be emphasised that the older mother is particularly at risk of having a Down's child and that the argument here is for the extension of the search for the disorder and not a shift of resources into another age grouping. Figure 2 demonstrates that vigilance is always required where the mother is older. The calculated risk rises to 0.84 per cent in the 35–39 age group, 2.31 per cent in the 40–44 age group and 4.87 per cent in those over 45 years (Ferguson-Smith, 1978). The evidence is so strong here that it ought to be normal practice for a general practitioner to discuss the possible diagnosis with a mother over the age of 35 years. There are good reasons for doing this with the mother before her referral to the obstetrician, so that she might be encouraged to accept advice concerning the diagnosis. Techniques which are less invasive, less costly and involve less delay in diagnosis than amniocentesis would be of great benefit in these cases.

There is, however, a much more important effect of the lowering of parental age in Down's syndrome and that is the lengthening of the span of years over which parents care for the Down's individual. Hagard and Carter (1976) estimated that the potential annual cost to the community of caring for a Down's individual was £4,150 at 1974 values. Included in their calculations were the cost of permanent care, education, loss of maternal income, the inability of Down's people to work and, therefore, the cost of their unemployability to society. Thus the individuals in this survey could cost up to £11.2 million (allowing for inflation) in the course of one year, depending on individual outcome. What is the cost, however, of the care which they do receive from parents—particularly from solitary mothers—as they grow older? It is apparent from this survey that parents and families see this care as a lifetime task. The proportion of Down's individuals who live either with only one parent or with relatives other than parents increases with age (Table 4), a pattern which causes concern as to whether this lifetime commitment is borne of choice or necessity.

The family doctor is in a unique position to study the morbidity of Down's individuals and their families. This survey was only the preliminary to a multipractice

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prospective study of 200 Down's children aged up to 10 years and control children matched for age, sex, family size and social class. Both parents and children will be studied with respect to their previous history and their morbidity during the year 1981. It is hoped that, using these considerable numbers of Down's individuals and their families through the observations of the family doctor, the natural history of the effect of mental handicap in families will be studied, not only in its early stages but into adulthood and particularly at the termination of the relationship through the death of the one parent, when it seems that most Down's individuals, having received a lifetime of individual family care, are decanted into institutions. Such studies might, hopefully, allow society to decide on objective grounds whether the Down's syndrome infant is a severe genetic malformation incompatible with family survival and therefore subject to therapeutic antenatal or post-natal termination, or an angel whom we entertain to the great enrichment of our society.

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Health Education Council budget 1982/3

The Council's budget is to be increased by more than £2 million to £8,572,000, chiefly because it is to include over £2 million especially provided for anti-smoking activities.

Source: Speech by Mr Geoffrey Finsberg, Parliamentary under Secretary of State at the Department of Health, 17 February 1982.

Childhood asthma

Asthmatic children with bronchial reactivity to a seasonal allergen, rye grass pollen and/or a perennial allergen, *Dermatophagoides pteronyssinus,* were studied over five years. Changes in their clinical symptoms and drug usage were correlated with changes in levels of serum IgG and IgE antibodies to the rye and mite allergens. The overall clinical severity of the children's asthma declined significantly during the study period and was accompanied by a significant fall in IgG and IgE anti-mite antibodies. The asthma of those children with bronchial reactivity to rye grass pollen continued to be exacerbated in the pollen season of year five, and both IgG and IgE anti-rye antibodies continued to rise and fall in relation to the season. These observations suggest that long-term perennial allergen exposure favours the induction of clinical and immunological hyporesponsiveness, whereas intermittent seasonal allergen exposure is associated with persistent clinical and immunological hypersensitivity.

Source: Hill, D. J., Hosking, C. S., Shelton, M. J. *et al.* (1981). Growing out of asthma: clinical and immunological changes over five years. *Lancet,* **2,** 1359-1362.