

Adults with Down's syndrome: the prevalence of complications and health care in the community

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ABSTRACT

Background

Individuals with Down's syndrome are predisposed to a variety of medical conditions which can impose an additional, but preventable, burden of secondary disability. Although there are guidelines for health checks and medical management of children with Down's syndrome, the needs of adults are relatively neglected.

Aim

To determine the prevalence of common medical problems in adults with Down's syndrome, and to assess current practice regarding medical surveillance of these patients.

Design of study

Detailed notes analysis.

Setting

Data were obtained from the primary care records of adults with Down's syndrome living in the Newcastle upon Tyne and Gateshead areas.

Method

Case notes were reviewed to obtain details regarding complications and to determine the frequency of medical surveillance of individuals with Down's syndrome.

Results

Complications such as hypothyroidism, coeliac disease, and obesity occur more frequently in adults with Down's syndrome than previous paediatric prevalence studies suggest. Surveillance of common complications that occur in individuals with Down's syndrome is infrequent. In this study, 48% of adults with Down's syndrome had not seen a doctor in the previous 12 months and 33% had not had a medical assessment in the previous 3 years.

Conclusion

Many individuals with Down's syndrome do not have access to regular healthcare checks, despite the high frequency of common medical complications in adult life. Debate regarding the practicality and relevance of introducing regular health checks is warranted.

Keywords

Down's syndrome; genetic screening; health care surveys; prevalence; screening.

INTRODUCTION

Down's syndrome is the most common identified cause of intellectual disabilities in the UK,¹ with a prevalence of 10.3 per 10 000 registered births.² Some medical conditions are over-represented in people with Down's syndrome, although accurate prevalence data are not widely published.³ Most of these conditions are treatable disorders which, if undiagnosed, impose an additional but preventable burden of secondary disability.

The Down's Syndrome Medical Interest Group (DSMIG)⁴ and the American Academy of Pediatrics⁵ developed guidelines for the medical management of children with Down's syndrome which provide surveillance recommendations. In practice, these guidelines are followed by paediatricians.⁶ However, the needs of adults have been relatively neglected in research and policy.

Provision of medical services for adults with Down's syndrome is becoming increasingly important. In the past 50 years, survival beyond the first year of life has improved remarkably for people with Down's syndrome: from below 50% to more than 90%.⁷ Median age at death has increased from 25 years in 1983 to 49 years in 1997, although there are significant differences between various racial and social groups.⁸ In recent years, the predicted

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Submitted: 9 January 2006; Editor's response: 16 March 2006; final acceptance: 4 August 2006.

©British Journal of General Practice 2007; 57: 50–55.

life expectancy has risen to 60 years.⁹ Of individuals with Down's syndrome recently born in the UK, 44% are expected to survive to the age of 60 years and 14% to 68 years (compared with 86% and 78% for the general population respectively).⁹ The number of individuals with Down's syndrome aged over 50 years has been predicted to increase by 200% between 1990 and 2010.¹⁰

The importance of identifying individuals with intellectual disabilities in primary care has also been highlighted in the 2006–2007 revision of the Quality and Outcomes Framework as part of the NHS General Medical Services contract.¹¹ Practices are now expected to produce a register of patients with intellectual disabilities.

The aims of this study were as follows:

- to determine the prevalence of common medical problems in adults with Down's syndrome; and
- to describe the level of care given to these adults living in a defined geographical area (Newcastle upon Tyne and Gateshead areas).

In this article the results of the review are discussed in relation to medical surveillance recommendations from the DSMIG⁴ and other sources.^{1,12–14}

METHOD

Data were obtained from the primary care records of adults with Down's syndrome living in the Newcastle and Gateshead area. Individuals were identified by community intellectual disability teams (including psychiatrists, nurses, physiotherapists, occupational therapists, psychologists, and speech and language therapists) and social service intellectual disability teams. Adults with intellectual disabilities are known by these teams through various routes including self-referral, duty referral, medical referral, and notification from social services after benefit claims.

GPs were contacted to gain permission to review primary care notes. After authorisation was obtained, anonymised information of medical problems was recorded from patients' notes. Information regarding screening or surveillance health checks undertaken since early adulthood was recorded. In particular, information was sought regarding cardiac disease, thyroid function, hearing and vision, coeliac disease, weight, and dementia.

After these data were available, they were examined in the context of recommendations by the DSMIG⁴ and others.^{1,12–14} The evidence for these recommendations is variable and they are not considered 'a blueprint for Gold Star services';⁴

How this fits in

This study demonstrates that many adult patients with Down's syndrome do not receive regular medical attention or health checks. It also demonstrates a high prevalence of several medical conditions which may result in an additional burden of secondary disability if untreated. This study suggests that there are potential benefits of establishing and disseminating an evidence-based guideline detailing structured health checks for adults with Down's syndrome.

however, they do provide a starting point for considering how structured health checks may be developed in the future for this population. Specific recommendations are presented with the results and their limitations are discussed.

A review of notes, conducted by researchers, took place between March and September 2004.

RESULTS

A total of 89 adults with Down's syndrome were known to intellectual disability services in the Newcastle upon Tyne and Gateshead areas. Permission was obtained to review the notes of 64 individuals (72%). Mean age of individuals whose notes were reviewed was 43.8 years (range = 18–61 years). Thirty-four (53%) of those whose notes were reviewed were female.

Medical complications

The prevalence of medical complications known to occur more frequently in individuals with Down's syndrome are shown in Table 1.

Medical review

Recommendation: Children with Down's syndrome should have an annual paediatric review.⁴ (There is no specific recommendation for medical review in adulthood.)

In total, 24 of 64 adults in this study (38%) had been seen by their GP in the previous 12 months. Of these individuals, 13 (20%) also received regular care from physicians based in secondary or tertiary care. A further nine individuals had not seen their GP in the previous 12 months but were receiving follow-up from hospital physicians for ongoing medical conditions.

Thirty-one (48%) individuals in this study had not seen any doctor in the previous 12 months; 24 (38%) individuals had not been medically reviewed in the previous 3 years.

Genetic investigations

Recommendation: The diagnosis of Down's syndrome should be confirmed by chromosome analysis.¹

Seventeen (27%) adults in this study had chromosome analysis. Records from the regional genetics centre were also checked to verify which individuals had undergone chromosome analysis. Of the individuals who had chromosome analysis, two had a Robertsonian t(14;21) translocation and the remainder had trisomy 21. One individual had not had a karyotype determined, despite having a family history of Down's syndrome.

Cardiac disease

Recommendation: Examination with a single echocardiogram should be performed in adult life,^{13,15} as there is an increased incidence of adult onset mitral valve prolapse and aortic regurgitation.¹⁶ In potential risk situations for infective endocarditis (for example, dental treatment and urogenital procedures), adults with Down's syndrome who have not had an adult echocardiogram should be given prophylactic antibiotics.¹³

Nine (14%) adults in this study had congenital heart disease. Five individuals had septal defects (one with patent ductus arteriosus), two patients

had tetralogy of Fallot, and two had congenital valvular disease.

All those with structural abnormality diagnosed in childhood had been informed of antibiotic cover according to records. However, in the absence of congenital heart disease no patient had an echocardiogram in adulthood. These individuals did not receive prophylactic antibiotics in potential risk situations for endocarditis.

Thyroid disorders

Recommendation: Biochemical testing should be carried out at least once every 2 years throughout life.^{17,18} Consideration of hypothyroidism is also mandatory in the differential diagnosis of depression and dementia.

Only 25 (39%) adults had their thyroid function checked in the last 2 years. Twenty-eight (44%) had not had a thyroid assessment in over 5 years.

Hearing impairment

Recommendation: Lifelong audiological surveillance is essential for individuals with Down's syndrome.¹⁹ Assessment of auditory thresholds, impedance testing, and otoscopy should be repeated at least once every 2 years. Hearing assessment is also essential in the differential diagnosis of depression and dementia.

Patients without diagnosed hearing difficulties had not undergone audiological surveillance. Patients diagnosed with depression and dementia had not had formal hearing assessment as part of a differential diagnosis.

Ophthalmic problems

Recommendation: Vision of individuals with Down's syndrome should be checked at least every 2 years throughout life.²⁰

The frequency of ophthalmic disorders is shown in Table 1. Details of the prevalence of refractive errors and optician surveillance were not available from medical notes.

Coeliac disease

Recommendation: Individuals should be clinically screened by history and examination on an annual basis to check for features that would prompt a blood test to check for antiendomysial antibody status.²¹ All those with existing thyroid disease, type 1 diabetes mellitus, or anaemia should be tested on a regular basis.

Patients had not been screened clinically on an annual basis and surveillance did not take place in individuals with thyroid disease or diabetes.

Weight

Recommendation: Appropriate guidance should be

Table 1. Medical problems in adults with Down's syndrome.

Medical problem	n (%)
Congenital heart disease	9 ^a (14)
Septal defect	5
Patent ductus arteriosus	1
Tetralogy of Fallot	2
Congenital valvular disease	2
Hypothyroidism	15 (23)
Coeliac disease	7 (11)
Type 1 diabetes mellitus	3 (5)
Seizures	18 (28)
Adult onset	12
Child onset	6
Depression	12 (19)
Alzheimer's disease	10 (16)
Hearing deficit	21 (33)
Ophthalmic disorders	23 (36)
Cataracts	8
Strabismus	4
Nystagmus	3
Glaucoma	2
Retinitis pigmentosa	2
Blindness	2
Corneal guttata	1
Keratoconus	1
Eczema	15 (23)
Osteoarthritis	9 (14)

^aPatients may have more than one medical problem associated with congenital heart disease.

given regarding diet. Exercises should be given to individuals with Down's syndrome, and thyroid function should be checked in those with accelerated weight gain.²²

Mean body mass index (BMI) of individuals in this study was 30.9 (range = 23.9–40.8). Thirteen (20%) had BMI greater than 35 and were therefore classed as obese. Only two (3%) patients were under review by a dietician. Fifteen of the 27 patients with obesity (56%) had not had thyroid assessment. There was no reference to dietary advice or weight control in patients' notes provided by members of the primary care teams.

Alzheimer's disease

Recommendation: Depression should be excluded before a diagnosis of Alzheimer's disease is made, as symptoms of depression mimic dementia.²³ Thyroid disorders and hearing loss should also be considered. Where there is doubt, brain imaging can be helpful.

Ten (16%) patients had a diagnosis of dementia (mean age = 52 years, range = 40–61 years). Of those individuals over the age of 40 years, 15/26 (58%) had a diagnosis of Alzheimer's disease.

Over a third of individuals with a diagnosis of Alzheimer's disease did not appear to have differential diagnoses (depression, hypothyroidism, and deafness) as a cause of symptoms excluded. No individual had brain imaging to confirm diagnosis.

Immunisations

Recommendation: All individuals with Down's syndrome should receive all vaccines according to the UK schedule: DTP (diphtheria, tetanus, and pertussis), polio, HiB (Haemophilus influenzae type b), Men C (Meningococcal C conjugate vaccines), MMR (measles, mumps, and rubella), and BCG (Bacille Calmette-Guérin). Influenza, pneumococcal, and hepatitis B vaccinations should also be considered.²⁴

All individuals had standard UK immunisations. However, only 28 (44%) had been immunised against influenza and 24 (38%) against pneumococcus; none were immunised against hepatitis B.

DISCUSSION

Summary of main findings

This study indicates a high prevalence of common, treatable medical conditions in adults with Down's syndrome. It also demonstrates a low level of medical involvement in these patients. Nearly half of the individuals studied had not seen a doctor 12 months before the study, and nearly a third had

not had a medical assessment in the previous 3 years. Surveillance is infrequent for common complications that occur in individuals with Down's syndrome and which can result in additional disability.

Strengths and the limitations of the study

A potential limitation to this community-based study is that it is unclear whether the cohort is representative of the population of individuals with Down's syndrome as a whole. This was inevitable because ascertainment in this study is likely to be high as multiple sources were used. Using an estimated number of individuals of all ages with Down's syndrome in the UK of 30 000,²⁵ the predicted number of adults with Down's syndrome in the Newcastle upon Tyne and Gateshead areas (population 320 000) is similar to the number that the research group successfully identified. The number is also similar to that found elsewhere, with 77 individuals with Down's syndrome over the age of 30 years identified in a population of 280 000 in the east of England.²³ Therefore, the researchers are confident that ascertainment in this study is representative of the adult Down's syndrome population.

Although it is possible that there may have been a non-response bias, it is unlikely that non-responders were receiving preferential medical treatment compared with responders.

A limitation of this study is that current practice was compared with a variety of recommendations from various documents and publications, rather than an accepted gold standard. This was inevitable because there is no widely accepted or disseminated standard of care for this group of patients. The absence of evidence-based guidance was evident to some of the GPs in this study. For example, one GP had been consulted by a family asking for continuing medical surveillance. The GP argued that surveillance guidelines for this population are confused and did not want to pursue specific tests without further expertise or evidence.

Some of the recommendations are straightforward. With regard to genetic testing, about 1 in 20 individuals with Down's syndrome have an unbalanced translocation rather than 'regular' trisomy 21.²⁶ In these individuals, there is a significant chance that parents and siblings carry a balanced translocation. Siblings of these adult individuals may be at a stage of planning to have children. To provide accurate information about the chances of them having children with Down's syndrome, it is essential to know the karyotype of the adults with Down's syndrome.

Other recommendations reflect a high prevalence of certain medical conditions. Single echocardiogram is recommended in view of studies showing that over half of adults with Down's syndrome may develop new structural abnormalities.¹⁶

However, other recommendations are more problematic. Some are drawn up by lobby groups and the evidence for specific recommendations is unclear. For instance, although immunisations have been advised²⁴ and brain imaging is thought to be helpful,²³ details regarding situations in which these are indicated are not clear.

There may also be concerns regarding the practicality of some recommendations. For instance, regular blood tests in adults with intellectual disability may be difficult. However, dried blood spot thyroid-stimulating hormone measurement is effective for detecting hypothyroidism in Down's syndrome, and capillary sampling with finger prick testing may be easier than taking a venous sample.⁶

The lack of disseminated, evidence-based guidelines means that clinicians looking after these patients cannot currently be expected to follow a single examination strategy. This study presents an analysis of current practice which can be used a benchmark when considering the development of future strategies.

Comparison with existing literature

This study reinforces earlier findings that demonstrate the presence of health problems in a large proportion of adult patients with intellectual disabilities.²⁷ Many of these problems are undiagnosed and health care for this population can be improved by the provision of structured health checks.

Such health checks already exist for paediatricians to ensure that children with Down's syndrome who develop Down's syndrome-related medical problems are diagnosed at an early stage and do not suffer from secondary disability.⁶ These programmes during childhood have been shown to be successful in diagnosing previously unrecognised health problems such as hypothyroidism⁶ and coeliac disease.²⁸

Implications for clinical practice

This study provides a starting point for discussion regarding what may constitute good care for adults with Down's syndrome. The development and dissemination of evidence-based national guidance detailing structured health checks for adults with Down's syndrome may help to standardise care. Guidance would help to identify evidence-based

screening procedures and determine their appropriate frequency.

One problem for those caring for adults with Down's syndrome is that it is unclear whose role it is to provide ongoing medical surveillance. Healthcare professionals who may be able to assist are psychiatrists, hospital physicians, clinical geneticists, and paediatricians (particularly at the time of transition from child health), as well as non-medical professionals. However, primary care is uniquely positioned to provide the comprehensive and coordinating role that is required. Therefore, the participation of GPs in debating the practicality and relevance of healthcare checks for adults with Down's syndrome is essential.

Ethics committee

Ethical approval for the project was obtained from the Newcastle and North Tyneside Local Research Ethics Committee (2003/126)

Competing interests

The authors have stated that there are none

Acknowledgements

We acknowledge the assistance and comments of Dr Judith M Rankin, University of Newcastle upon Tyne, during the analysis of results and preparation of this article. We are also grateful for the assistance of psychiatrists, physicians, psychologists, physiotherapist, occupational therapists, and speech and language therapists in Newcastle and Gateshead as well as adult individuals with Down's syndrome in the region.

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