Routledge
Taylor & Francis Group

Journal of Intellectual and Developmental Disability

ISSN: 1366-8250 (Print) 1469-9532 (Online) Journal homepage: http://www.tandfonline.com/loi/cjid20

Healthcare and guidelines: A population-based survey of recorded medical problems and health surveillance for people with Down syndrome

Tuomo Määttä, Joonas Määttä, Tuula Tervo-Määttä, Anja Taanila, Markus Kaski & Matti Iivanainen

To cite this article: Tuomo Määttä, Joonas Määttä, Tuula Tervo-Määttä, Anja Taanila, Markus Kaski & Matti Iivanainen (2011) Healthcare and guidelines: A population-based survey of recorded medical problems and health surveillance for people with Down syndrome, Journal of Intellectual and Developmental Disability, 36:2, 118-126

To link to this article: http://dx.doi.org/10.1080/13668250.2011.570253

Journal of Intellectual & Developmental Disability, June 2011; 36(2): 118–126

informa
healthcare

Healthcare and guidelines: A population-based survey of recorded medical problems and health surveillance for people with Down syndrome

TUOMO MÄÄTTÄ[1], JOONAS MÄÄTTÄ[2], TUULA TERVO-MÄÄTTÄ[3], ANJA TAANILA[4,5], MARKUS KASKI[6] & MATTI IIVANAINEN[6,7]

[1]*Service Centre of Kuusanmäki, Finland,* [2]*Medical Faculty, University of Oulu, Finland,* [3]*Department of Teacher Education, University of Oulu, Kajaani, Finland,* [4]*Unit of General Practice, University Hospital of Oulu, Finland,* [5]*Institute of Health Sciences, University of Oulu, Finland,* [6]*Rinnekoti Research Centre, Espoo, Finland, and* [7]*Department of Child Neurology, University of Helsinki, Finland*

Abstract

Background Medical problems are described in a population of persons with Down syndrome. Health surveillance is compared to the recommendations of national guidelines.

Method Case records from the specialised and primary healthcare and disability services were analysed.

Results A wide spectrum of age-specific medical and surgical problems was described. Congenital heart defects and middle ear infections were mostly experienced by younger people, while thyroid disease, epilepsy, and Alzheimer's disease were frequent among older people. Psychiatric disorders and behavioural problems were frequent in all age groups.

Conclusions Health surveillance remained insufficient, despite the guidelines available. A joint effort by healthcare and disability service providers is required to ensure that the medical needs of people with Down syndrome are adequately met across their entire lifespan. An active provision of healthcare and monitoring for this vulnerable group is needed.

Keywords: *Down syndrome, morbidity, healthcare, health surveillance, guidelines, case record study*

Introduction

The birth prevalence of people with Down syndrome has remained relatively stable during the last decade (Collins, Muggli, Riley, Palma, & Halliday, 2008) and their survival has improved (Weijerman et al., 2008). Their life expectancy has increased to nearly 60 years of age during the last two generations in developed countries (Janicki, Dalton, Henderson, & Davidson, 1999; Merrick, 2000). Nevertheless, on average, individuals with Down syndrome die at an earlier age than do adults with intellectual disability and adults in the general population (Bittles, Bower, Hussain, & Glasson, 2007; Janicki et al., 1999). Furthermore, the rates of reported medical issues are high in clinical samples among children and teenagers (Yam et al., 2008) and adults (Henderson, Lynch, Wilkinson, & Hunter, 2007; Kerins, Petrovic, Bruder, & Gruman, 2008), as well as in population-based health studies (Murphy et al., 2005; Schieve, Boulet, Boyle, Rasmussen, & Schendel, 2009).

Congenital heart defects occur in half of all children born with Down syndrome, and the mortality of people with severe heart defects was high before modern heart surgery (Hijii, Fukushige, Igarashi, Takahashi, & Ueda, 1997). Hospital admission and medication use rates in young infants with Down syndrome are still very high, mainly because of congenital heart and gastrointestinal diseases and respiratory infections (van Trotsenburg, Heymans, Tijssen, Vijlder, & Vulsma, 2006).

Visual impairment (Creavin & Brown, 2009; Stephen, Dickson, Kindley, Scott, & Charleton,

Correspondence: Tuomo Määttä, Chief Physician, Joint Authority for Kainuu, Service Centre of Kuusanmäki, 87250 Kajaani, Finland.
E-mail: tuomo.maatta@kainuu.fi

ISSN 1366-8250 print/ISSN 1469-9532 online
DOI: 10.1080/13668250.2011.570253

2007) and hearing loss (Buchanan, 1990; Meuwese-Jongejeugd et al., 2006; van Schrojenstein Lantman-de Valk et al., 1994) are frequent among people with Down syndrome at all ages. Methods for hearing and visual acuity assessments that are used for the mainstream population are often inadequate for people with intellectual disability. Organised visual and hearing assessments using appropriate methods are urgently needed (van Schrojenstein Lantman-de Valk et al., 1994) but are still difficult to implement (Evenhuis et al., 2004). Indeed, continuous ophthalmological monitoring is crucial (Creavin & Brown, 2009; Jönelid, Annerén, & Holmström, 2002). Establishment of ophthalmic screening improves ocular surveillance, giving hope of improved developmental and functional outcomes in individuals with Down syndrome (Stephen et al., 2007).

As a group, infants with Down syndrome have a persistent mild congenital hypothyroidism (van Trotsenburg, Kempers, et al., 2006). The prevalence of overt thyroid disease increases to 7% in children (Gibson et al., 2005; Murphy et al., 2008; Unachak et al., 2008) and further during adulthood. In adults, the 10-year incidence of definite hypothyroidism is 13.6% (Prasher & Gomez, 2007). Screening for hypothyroidism annually has been recommended for children (American Academy of Pediatrics, 2001) and every one to two years for adults. Guidelines have increased detection and treatment of thyroid disease (Carroll, Arbogast, Dudley, & Cooper, 2008).

The prevalence of coeliac disease is 6–8% among children and adolescents (Carnicer et al., 2001; Nisihara et al., 2005; Sciberras, Vella, & Grech, 2004). Screening all children with Down syndrome for coeliac disease has been recommended (Van Cleve & Cohen, 2006), however, the cost-effectiveness of this screening has been questioned (Swigonski, Kuhlenschmidt, Bull, Corkins, & Downs, 2006).

Depression and behavioural disorders are frequent (Dykens, 2007; Määttä, Tervo-Määttä, Taanila, Kaski, & Iivanainen, 2006) and in adults these may precede the onset of dementia (Burt, Loveland, & Lewis, 1992; Dykens, 2007; Urv, Zigman, & Silverman, 2008). Epilepsy and declining adaptive behaviour are common in ageing people with Down syndrome (Collacott, 1993). People with Down syndrome have a high risk for early Alzheimer's disease and premature ageing (Coppus et al., 2006; Zigman & Lott, 2007). The comorbidity of Down syndrome and Alzheimer's disease is an increased reality due to an increased life expectancy and the very early onset of Alzheimer's disease in this population. There is some evidence of the benefit of medical treatment on the outcome of Alzheimer's disease (Prasher, Huxley, Haque, & the Down syndrome Ageing Study Group, 2002), but larger randomised controlled studies with longer follow-ups are required. It is crucial to recognise and treat all health comorbidities in ageing persons with Down syndrome and Alzheimer's disease (Kerins et al., 2008; McCarron, Gill, McCallion, & Begley, 2005).

Many individuals with Down syndrome do not have access to regular healthcare checks, despite the high frequency of medical complications (Henderson et al., 2007). Comprehensive health assessment programs have improved health outcomes among people with intellectual disability (Cooper et al., 2006; Lennox et al., 2007). Health-check intervention of adults with intellectual disability has improved the health and reduced costs of care compared with standard care (Romeo et al., 2009). Low adherence to national healthcare guidelines for people with Down syndrome has recently been reported (Fergeson et al., 2009; Virji-Babul, Eichmann, Kisly, Down, & Haslam, 2007).

National recommendations for good healthcare for people with Down syndrome were developed for health professionals of Finland (Kaski et al., 2004) and are easily available online for physicians, along with other evidence-based Current Care[1] guidelines, for use during consultations. According to the Finnish guideline, an ophthalmologist should evaluate vision at the age of six months, at the start of school and commencement of work, and then regularly throughout the lifespan. Hearing should be tested yearly throughout childhood and every three years during adulthood. Thyroid function should be monitored every two years throughout life. Screening for coeliac disease in the presence of even slight symptoms is recommended at any age.

Our aim was to describe health concerns and care recorded in case records of people with Down syndrome in the population of the Kainuu region, Finland. The recent state of health surveillance was compared to the national guidelines. The specific aims of the survey were to describe the frequencies of

(1) diagnosed impairments and diseases (e.g., congenital and acquired heart defects and common diseases) throughout the lifespan,
(2) treatments (e.g., surgery of observed heart defects and other conditions with operative treatment options, use of specific diets), and
(3) selected efforts to detect diseases (e.g., tests of thyroid and coeliac disease).

Study population

A population-based study of case records of people with Down syndrome was performed. One hundred and thirty-seven persons with Down syndrome were identified in the Intellectual Disability Service Register of the Kainuu region in Northern Finland, and they were all included in the survey.

In December 2008, the population of the Kainuu region was 79,690; the number of people with intellectual disability living in this region was 723, according to the disability service registers. Eighty-four people identified as having Down syndrome were living in the region. Thus, the prevalence of people identified with Down syndrome was 1.05/1,000, and 11.6% among the population with intellectual disability. Disability and health register data were available for 32 deceased people who had had Down syndrome, and for 21 people who had lived in the region and moved elsewhere. The age distribution of the people with Down syndrome in this study is given in Table 1.

A chromosome analysis was available for 113 participants (82%). Trisomy of chromosome 21 had been confirmed in 108 (96%) of these cases. Translocation trisomy had been found in three participants, and mosaic trisomy in two participants.

The severity of intellectual disability was mild to moderate in 49% and severe to profound in 51% of people who had had cognitive evaluations. Twenty-two percent of the people with Down syndrome used nonverbal communication only, 35% used single word utterances, and 43% were able to produce at least short spoken sentences.

Methods

The Ethics Committee of the Central Hospital of Kainuu approved the study. The Ministry of Social and Health Affairs gave permission for combining data from medical and disability service records.

The case records of these people with Down syndrome were analysed for both specialised and primary healthcare and disability services. Data collection from the archives of the Intellectual Disability Service Register and the Central Hospital of Kainuu was performed in March 2004, and updated during January 2009. Original case record data of these registers from birth to the data collection date were surveyed. The second review of case records was conducted online from the regional computerised case records that were used for at least five years in all public primary and specialised health and disability services. The data of health surveillance, primary healthcare, and dental care were included. Laboratory examinations performed during the preceding five years were reviewed. The combined data of these two surveys was used for analysis. The data of persons who moved from the region were analysed for the time they lived in the region and in the local registers only. The first author, a physician with expertise in the field of intellectual disability, surveyed the recorded data.

The detected health impairments were related to two age groups: 0–29 years, and 30 years or older. The first group included children and young adults; the second represented middle-aged and older participants. The two age groups were compared to ascertain age-related differences in recorded health issues. The older group born 30 years ago or more did not have access to treatments such as surgery for severe heart defects that were available to most in the younger age group.

The combined data of deceased and relocated people is presented separately in the results as a past subgroup ($n = 53$). The age distribution in the subgroups of participants is given in Table 1. The data of the past subgroup were compared to the present population ($n = 84$) to detect possible recent changes in recorded health problems, care, and health surveillance. The characteristics of the past and present subgroups are given in Table 2. The time from the last available recorded data was 10 years longer in the past compared to the present subgroup.

Table 1. Age distribution of people with Down syndrome in the study

Age	Present		Deceased		Relocated		All	
	n	%	*n*	%	*n*	%	*n*	%
0–9	11	13	3	9	6	29	21	15
10–19	10	12	1	3	2	9	12	9
20–29	11	13	3	9	3	14	18	13
30–39	16	19	5	16	4	19	24	17
40–49	20	24	5	16	5	24	31	23
50–59	16	19	8	25	1	5	26	19
60+	0	0	7	22	0	0	5	4
Total	84	100	32	100	21	100	137	100

Health services for the study population

Everyone residing in Finland is entitled to receive good quality public healthcare within set time frames. Public health services are divided into primary health, specialised medical, and hospital care. Private healthcare supplements public health

Table 2. Characteristics of the past (deceased or re-located) and present subgroups

Characteristic of the group	Past	Present
	$n = 53$	$n = 84$
Males, *n* (%)	28 (52)	49 (59)
Age years, mean (range)	35.1 (0.1–67.9)	34.5 (0.6–59.2)
Age group of 0–29 years, *n* (%)	20 (38)	30 (36)
30-year-old age group, *n* (%)	33 (62)	54 (64)
Time from last contact or death, years (range)	13.5 (0.3–34)	3.6 (0.1–23.6)
Deceased participants, *n* (mean age at death)	32 (43.3)	0
Participants no longer in target region, *n* (mean age at last contact)	21 (22.7)	0

services. Special services for people with intellectual disability, covered by a variety of legislation, are provided as positive special treatment to safeguard equality.

The *specialised healthcare/Central Hospital* provide a complete paediatric evaluation of all children born with Down syndrome, including chromosome analysis, ultrasounds of the heart, thyroid function assessments, and hearing assessments by otoacoustic emission after birth. Specialists evaluate the heart defects and other severe health impairments. However, there is no systematic follow-up program after the neonatal period in the specialised healthcare for these children. All children born with Down syndrome have been referred to regional disability services soon after birth during the last three decades.

The *disability services/Service Centre* provide(s) developmental and health assessments to all people with Down syndrome from early childhood to adulthood. Adults are met by the expert team of the specialised disability services only when deemed necessary. However, anxiety, depression, dementia, and behavioural symptoms are often assessed by the first author working in this team. Psychiatric consultations are used for severe psychiatric symptoms.

The *primary healthcare/Health Centre* is responsible for preventive healthcare (e.g., regular assessments of hearing and vision problems in child welfare clinics and school healthcare), first aid (e.g., assessment of pain, accidents, fainting, and convulsions, and treatment of respiratory, urinary, and skin infections), and for following up on hypothyroidism and other chronic diseases. General practitioners send patients to other specialists for consultations when deemed necessary.

Private specialists are available too, but are seldom used by the study population in comparison to their use of public services. However, ophthalmologists and optometrists provide services to correct mild-to-moderate refractive errors for patients without eye diseases or severe disability, preferably in the private sector.

Occupational healthcare is the primary form of preventive healthcare for employees. Occupational healthcare services are seldom available for people with intellectual disability.

Each of the health and social service providers described in the previous paragraphs uses separate case records and registers. Even within specialised healthcare, somatic and mental health services have their own case records. The permission given by the client or his/her stakeholder is required for any client data referral between the registers.

Results

The detected health impairments were related to two age groups (0–29 years, and 30 or more years) and two subgroups (present and past members) of the study population. The rates of selected common medical problems as percentages of affected people in each group are given in Table 3. Surgical treatments, diets, and screenings are summarised in Table 4.

The majority of visits by adults to medical services (including visits to GPs and specialists in Central Hospital) related to assessments of symptoms and follow-up of detected medical problems. Comprehensive preventive health assessments of adult participants were infrequently reported.

Heart and circulation

Congenital heart defects were more frequent among younger people (48%) than among older people (15%). Diastolic dysfunction with poor relaxation and impaired filling of the heart was seen in some adults with structurally normal hearts.

Ear infections, hearing loss

Middle ear infections were significantly more common in the younger participants (52%) than in the older participants and in the present compared to the past subgroup. Hearing loss was reported in 22% of the participants. Pure tone audiometry to accurately determine hearing loss was seldom performed successfully. An assessment of hearing using audiometry or otoacoustic emission had been recorded in 35% of participants in the present population, more often in younger participants (59%) than in the older participants (19%). Four of the 84 participants (5%) in the present population used hearing aids.

Table 3. Reported medical problems of people with Down syndrome in the Kainuu region

	Age groups			Groups			
	0–29 years $n = 50$	30 years and older $n = 87$		Past $n = 53$	Present $n = 84$		All $N = 137$
Medical problems	%	%	p	%	%	p	%
Congenital heart defects	48	15	***	28	26		27
Middle ear infections	52	20	***	11	44	***	31
Hearing loss	16	25		13	27		22
Eye concerns	50	67	***	45	70	**	61
Cataracts	6	22	**	17	16		16
Skin disease	10	17		15	14		15
Asthma	12	7		19	2	**	9
Hypothyroidism	8	33	**	21	26		24
Psychiatric or behavioural problem	22	34		19	37	*	30
Depression and/or anxiety	16	29		17	29		24
Challenging behaviour	10	23		9	24	*	18
Autistic features	6	9		8	8		8
Panic	0	9	*	0	10	*	8
Seizures, fits, panic, or fainting	34	52	*	49	49		45
Fainting	14	21		19	18		18
Epilepsy	4	21	**	26	7	**	15
Other emergencies							
Pneumonia	40	31		45	27	*	34
Stroke	4	17	*	28	2	***	12
Alzheimer's disease	0	34	***	28	18		22

*$p < .05$. **$p < .01$. ***$p < .001$. Pearson's chi-square test; Fisher's exact test was used when cells of the tables had values < 5.

Table 4. Treatments and diagnostics for people with Down syndrome in the Kainuu region

	Age groups			Groups			
	0–29 years $n = 50$	30 years and older $n = 87$		Past $n = 53$	Present $n = 84$		All $N = 137$
Intervention	%	%	p	%	%	p	%
Middle ear infections, tympanostomy (grommets)	24	7	**	4	19	*	13
Congenital heart defects							
Heart defect operated	18	3	**	4	12		9
Heart defect inoperable	14	6		19	2	**	9
Heart surgery not needed	16	6		7	12		9
Gastrointestinal surgery	12	21		23	14		18
Orthopedic surgery	4	20	**	9	17		14
Genitourinary surgery	8	11		17	6	*	10
Operated	36	55	*	36	56	*	48
Diet (because of food allergy, diabetes, or coeliac disease)	18	8		15	10		12
Thyroid test	10	64	***	30	54	**	45
Coeliac lab	2	18	**	2	19	**	12

*$p < .05$. **$p < .01$. ***$p < .001$. Pearson's chi-square test; Fisher's exact test was used when cells of the tables had values < 5.

Visual impairment

Seventy percent of the present population had eye concerns, including refractive errors and eye diseases. Eye concerns were significantly more common in the older participants (67%) compared to younger participants (50%), and had been recorded significantly more often in the present compared to the past subgroup. Glasses to correct refractive errors were widely distributed. Cataracts were detected in 6% of the young, and in 22% of the older group.

Mental health and behaviour

Psychiatric disorders or challenging behaviour were recorded in 30% of participants. A specific diagnosis was not easily achieved. Depression was sometimes diagnosed only after long and elaborate investigations. Maladaptive challenging behaviour was often an early sign of Alzheimer's disease.

Seizures, fits, and fainting

Acute loss or disturbance of consciousness had affected 45% of the participants. Fainting was the most common diagnosis, and was seen in 18% of participants. Acute anxiety symptoms, hyperventilation, and panic attacks mimicked epileptic seizures. Epilepsy was diagnosed in 21% of the ageing adults. Long, intractable seizures sometimes proved non-epileptic.

Other emergencies

Forty-seven (34%) participants had pneumonia at least once and 25 (18%) had repeated pneumonias, some because of aspiration. Strokes, including repeated strokes, had caused permanent disabilities for some of the young and many of the older participants.

Alzheimer's disease

Dementia was diagnosed only in persons older than 30 years: in 15 persons (28%) of the 54 in the present population. Medication for Alzheimer's disease was used by nine of 15 (60%) in the present population.

Other health concerns

Two children had acute leukaemia. Three participants lost their mobility because of spinal cord injuries; two of them had anomalies of the upper cervical spine, and the third had degenerative changes of the middle part of the cervical spine. Diabetes was diagnosed in five participants; three were treated with insulin. Skin infections proceeded repeatedly to abscesses in many patients. Asthma, psoriasis, and alopecia were seen in many patients. Fractures occurred in 12 participants.

Oral health

Fifty percent of the present population—78% of the younger (< 30 years) participants and 35% of the older (>30 years) participants—had visited a dentist or oral hygienist during the last three years. Within these two groups, 50% of the younger participants and 19% of the older participants had visited during the last year. The oral health of the younger participants was satisfactory; many had full or cured dentition. Chronic periodontal infection was the main concern and cause of dental loss during adulthood. Corrective dental care was performed under general anaesthesia for 44 (32%) of the participants, whose fearfulness prevented the usual care.

Surgical treatments

Forty seven (56%) of the currently living participants had at least one major surgical operation, and 37 (27%) of all participants had had several operations, even when adenoidectomies, tonsillectomies, grommet insertions, ophthalmologic and dental operations were not counted. Common indications for operative treatments were musculoskeletal (e.g., habitual dislocations of the knee, fractures, various hernias), gastrointestinal (e.g., gall stones, which were operated on in five of the nine detected, and duodenal obstructive anomalies), cardiovascular problems (e.g., congenital heart defects), and genitourinary problems (e.g., foreskin and other urinary tract strictures in males).

Screening for hypothyroidism and search for coeliac disease

Finnish health guidelines recommend thyroid tests every two years throughout life. Nevertheless, thyroid-stimulating hormone (TSH) determination had been performed for only 54% of the participants in the present population during the preceding five years. Hypothyroidism had been diagnosed in 24%; it was more common in older (33%) than younger (8%) participants. Laboratory examinations to detect coeliac disease were recommended when even the slightest suspicion arose. Coeliac disease had been diagnosed in three (4%) of the participants, and screening tests had been performed for 16 (19%) of the participants in the present population. Significantly, more participants in the present and older subgroups had participated in screenings for hypothyroidism and coeliac disease than in the deceased or relocated, and young subgroups.

Discussion

The two main aims of this study were to describe the detected medical problems and treatments in a regional population of people with Down syndrome,

and to compare their health surveillance to national guidelines.

All 137 people with Down syndrome identified in the Intellectual Disability Service Register in the Kainuu region of Northern Finland were included, and their health and disability case records in the public services analysed. Data for the majority of lifespans could be comprehensively surveyed using the original paper records of the disability and specialised healthcare archives. The case records of the primary healthcare and laboratory results from all public service producers were available online and reviewed for the preceding five years. Reliability of the survey of recorded data was not tested. The first author surveyed the recorded data. The same person repeated the survey of the records to update for current health information and to enhance reliability. In this paper, we describe only those health concerns that the databases allow and only those reported, not necessarily all those that are relevant to people with Down syndrome.

Inclusion of people who had died or had moved out of the area in the analysis of age-related health concerns and combining data of these different groups is problematic to the interpretation of the results. Nevertheless, we chose to do this in order to attain a sufficient number of participants for description of the most common medical problems. People who have died are likely to have had more health problems than those who have moved away based on their health status at those times. Inclusion of deceased people added information of severe medical problems and emergencies in all age groups. Specifically, heart defects, pneumonia, epilepsy, and stroke could be better described as a consequence.

Data on health services provided to this population by private specialists, including private ophthalmologists, were not available for the study. Thus, the extent of consultations of private ophthalmologists and assessments by optometrists for glass correction of refractive errors remained unknown. Data were not recorded to confirm that hearing acuity of adults and ageing people of this population was regularly assessed. For assessment of hearing loss there were no alternative services for the services that were surveyed in this study.

The data that were not reviewed or recorded may be just as important. However, the public health and disability services reviewed in this study are the health services accessible to all participants and the most commonly used services. As a rule, detected health problems and interventions are carefully recorded in the case records. Thus, the diseases identified and recorded in the analysed case records probably reflect identified medical problems and interventions undertaken. The findings reflect local clinical services and do not necessarily generalise to other populations and service arrangements.

Health problems were extensive from birth to old age, and many health concerns were age-specific. Older people with Down syndrome were definitely a distinct group, with few survivors among those with severe congenital heart defects. This may in part be because heart surgery has been available for less than 40 years. The authors acknowledge that the health needs of young children, youth, adults, and ageing people are far more complex than could be described in this study.

The differences in the morbidity, healthcare, and health surveillance between the present and past subgroups may to some degree reflect recent changes of health services, because there was a 10-year mean difference between the subgroups in the time since the last recorded data. In addition, the differences in the availability of data and the morbidities of the deceased participants in the past subgroup may contribute to the group differences.

There was lack of evidence that the healthcare guidelines were being followed during the last five years, since the guidelines were implemented for detection of thyroid disorder and hearing loss.

The main reasons for infrequent health surveillance might be that the need was not recognised by the patients, their proxies, or professionals. Physicians were informed about new Current Care guidelines in their main national medical journal *Duodecim* and as a link at their widely used website. Furthermore, regular health screening was not actively organised and offered. The guidelines were available to healthcare professionals and supported by many, but probably used as clinical guidelines only, rather than for planning and organising services. The clients, their proxies, and the service producers were perhaps not aware of the recommendations to participate in regular health screening. It is probably unrealistic to expect that advice from guidelines would be put into practice within a relatively short period of time, particularly as no information campaign was conducted.

To overcome the challenges related to the health of these patients, proxies, professionals, and even organisations have to communicate effectively with each other. One obvious communication barrier was between health and disability services with separate client registers. Responsibilities for health surveillance and care of children and adults were shared, but the partners seemed to be partly unaware of each other's practices. Consequently, children with Down syndrome did not have thyroid screenings as recommended. Shared patient records for health (primary, specialised, physical, and mental health) and disability services (local and regional) could improve the coordination of services.

The Finnish guideline's recommendation to screen for specific health concerns of people with Down syndrome did not guarantee these services in the population studied. A major joint effort of disability and health service providers is required to ensure that the medical needs of people with Down syndrome are adequately met across their entire lifespan. An active program of health monitoring seems to be essential to improving the health of people with Down syndrome.

Conclusions

People with Down syndrome have high risks of heart defects, visual impairment, hearing loss, depression, infections, hypothyroidism, epilepsy, and Alzheimer's disease. Sufficient healthcare improves their health and well-being. Based on the research literature and the present results, we conclude that

(1) health surveillance should be actively offered to people with Down syndrome throughout their lives,
(2) preventive health screening and diagnostic practices need to be developed and applied, and
(3) responsibilities need to be clearly defined and reported to health and disability service providers and clients to successfully implement guidelines of health surveillance.

Author note

The study was partly supported by the Joint Authority of Kainuu, the Rinnekoti Research Foundation, and the Finnish Cultural Foundation. They played no role in study design; collection, analysis, and interpretation of data; writing of the paper; and decision to submit it for publication.

Conflicts of interest: None.

Note

1 *Current Care* (Käypä Hoito) is a Finnish project producing evidence-based treatment guidelines for the Finnish Medical Society *Duodecim*. These guidelines are drawn up in support of healthcare professionals and for the benefit of patients (http://www.kaypahoito.fi/web/english/home).

References

American Academy of Pediatrics. Committee on Genetics. (2001). American Academy of Pediatrics: Health supervision for children with Down syndrome. *Pediatrics*, *107*, 442–449. doi:10.1542/peds.107.2.442

Bittles, A.H., Bower, C., Hussain, R., & Glasson, E.J. (2007). The four ages of Down syndrome. *European Journal of Public Health*, *17*, 221–225. doi:10.1093/eurpub/ckl103

Buchanan, L.H. (1990). Early onset of presbyacusis in Down syndrome. *Scandinavian Audiology*, *19*, 103–110. doi:10.3109/01050399009070760

Burt, D.B., Loveland, K.A., & Lewis, K.R. (1992). Depression and the onset of dementia in adults with mental retardation. *American Journal on Mental Retardation*, *96*, 502–511.

Carnicer, J., Farré, C., Varea, V., Vilar, P., Moreno, J., & Artigas, J. (2001). Prevalence of coeliac disease in Down's syndrome. *European Journal of Gastroenterology & Hepatology*, *13*, 263–267. doi:10.1097/00042737-200103000-00008

Carroll, K.N., Arbogast, P.G., Dudley, J.A., & Cooper, W.O. (2008). Increase in incidence of medically treated thyroid disease in children with Down syndrome after rerelease of American Academy of Pediatrics health supervision guidelines. *Pediatrics*, *122*, e493–498. doi:10.1542/peds.2007-3252

Collacott, R.A. (1993). Epilepsy, dementia and adaptive behaviour in Down's syndrome. *Journal of Intellectual Disability Research*, *37*, 153–160.

Collins, V.R., Muggli, E.E., Riley, M., Palma, S., & Halliday, J.L. (2008). Is Down syndrome a disappearing birth defect? *The Journal of Pediatrics*, *152*, 20–24. e1. doi:10.1016/j.jpeds.2007.07.045

Cooper, S.-A., Morrison, J., Melville, C., Finlayson, J., Allan, L., Martin, G., & Robinson, N. (2006). Improving the health of people with intellectual disabilities: Outcomes of a health screening programme after 1 year. *Journal of Intellectual Disability Research*, *50*, 667–677. doi:10.1111/j.1365-2788.2006.00824.x

Coppus, A., Evenhuis, H., Verberne, G.-J., Visser, F., van Gool, P., Eikelenboom, P., & van Duijin, C. (2006). Dementia and mortality in persons with Down's syndrome. *Journal of Intellectual Disability Research*, *50*, 768–777. doi:10.1111/j.1365-2788.2006.00842.x

Creavin, A.L., & Brown, R.D. (2009). Ophthalmic abnormalities in children with Down syndrome. *Journal of Pediatric Ophthalmology and Strabismus*, *46*, 76–82. doi:10.3928/01913913-20090301-06

Dykens, E.M. (2007). Psychiatric and behavioral disorders in persons with Down syndrome. *Mental Retardation and Developmental Disabilities Research Reviews*, *13*, 272–278. doi:10.1002/mrdd.20159

Evenhuis, H., van Splunder, J., Vink, M., Weerdenburg, C., van Zanten, B., & Stilma, J. (2004). Obstacles in large-scale epidemiological assessment of sensory impairments in a Dutch population with intellectual disabilities. *Journal of Intellectual Disability Research*, *48*, 708–718. doi:10.1111/j.1365-2788.2003.00562.x

Fergeson, M.A., Mulvihill, J.J., Schaefer, G.B., DeHaai, K.A., Piatt, J., Combs, K., ... Neas, B.R. (2009). Low adherence to national guidelines for thyroid screening in Down syndrome. *Genetics in Medicine*, *11*, 548–551. doi: 10.1097/GIM.0b013e3181a9c250

Gibson, P.A., Newton, R.W., Selby, K., Price, D.A., Leyland, K., & Addison, G.M. (2005). Longitudinal study of thyroid function in Down's syndrome in the first two decades. *Archives of Disease in Childhood*, *90*, 574–578. doi:10.1136/adc.2004.049536

Henderson, A., Lynch, S.A., Wilkinson, S., & Hunter, M. (2007). Adults with Down's syndrome: The prevalence of complications and health care in the community. *British Journal of General Practice*, *57*, 50–55.

Hijii, T., Fukushige, J., Igarashi, H., Takahashi, N., & Ueda, K. (1997). Life expectancy and social adaptation in individuals with Down syndrome with and without surgery for congenital heart disease. *Clinical Pediatrics*, *36*, 327–332. doi:10.1177/000992289703600603

Janicki, M.P., Dalton, A.J., Henderson, C.M., & Davidson, P.W. (1999). Mortality and morbidity among older adults with intellectual disability: Health services considerations. *Disability and Rehabilitation*, *21*, 284–294. doi:10.1080/096382899297710

Jönelid, B., Annerén, G., & Holmström, G. (2002). Children and adolescents with Down syndrome. Continuous ophthalmological monitoring crucial! [Barn och ungdomar med Downs syndrom. Ogonuppfoljning maste ske kontinuerligt!]. *Läkartidningen*, *99*(1–2), 29–32.

Kaski, M., Aaltonen, S., Autti-Rämö, I., Heiskanen, K., Huovinen, K., Hyvärinen, L., … Wilska, M. (2004). *Management of medical problems associated with Down Syndrome (Article ID: ccs00007, hoi50027 [050.027])*. Retrieved from http://www.kaypahoito.fi/

Kerins, G., Petrovic, K., Bruder, M.B., & Gruman, C. (2008). Medical conditions and medication use in adults with Down syndrome: A descriptive analysis. *Down Syndrome Research & Practice*, *12*, 141–147.

Lennox, N., Bain, C., Rey-Conde, T., Purdie, D., Bush, R., & Pandeya, N. (2007). Effects of a comprehensive health assessment programme for Australian adults with intellectual disability: A cluster randomized trial. *International Journal of Epidemiology*, *36*, 139–146. doi:10.1093/ije/dyl254

Määttä, T., Tervo-Määttä, T., Taanila, A., Kaski, M., & Iivanainen, M. (2006). Mental health, behaviour and intellectual abilities of people with Down syndrome. *Down Syndrome Research & Practice*, *11*, 37–43.

McCarron, M., Gill, M., McCallion, P., & Begley, C. (2005). Health co-morbidities in ageing persons with Down syndrome and Alzheimer's dementia. *Journal of Intellectual Disability Research*, *49*, 560–566. doi:10.1111/j.1365-2788.2005.00704.x

Merrick, J. (2000). Incidence and mortality of Down syndrome. *The Israel Medical Association Journal*, *2*, 25–26.

Meuwese-Jongejeugd, A., Vink, M., van Zanten, B., Verschuure, H., Eichhorn, E., & Koopman, D., … Evenhuis, H. (2006). Prevalence of hearing loss in 1598 adults with an intellectual disability: Cross-sectional population based study. *International Journal of Audiology 45*, 660–669. doi:10.1080/14992020600920812

Murphy, J., Hoey, H.M.C. V., O'Regan, M., O'Keeffe, M., Philip, M., Fogarty, E., … McShane, D. (2005). Guidelines for the medical management of Irish children and adolescents with Down syndrome. *Irish Medical Journal*, *98*, 48–52.

Murphy, J., Philip, M., Macken, S., Meehan, J., Roche, E., Mayne, P.D., … Hoey, H.M.C.V. (2008). Thyroid dysfunction in Down's syndrome and screening for hypothyroidism in children and adolescents using capillary TSH measurement. *Journal of Pediatric Endocrinology & Metabolism*, *21*, 155–163.

Nisihara, R.M., Kotze, L.M.S., Utiyama, S.R.R., Oliveira, N.P., Fiedler, P.T., & Messias-Reason, I.T. (2005). Celiac disease in children and adolescents with Down syndrome. *Jornal de Pediatria*, *81*, 373–376. doi:10.2223/JPED.1381

Prasher, V., & Gomez, G. (2007). Natural history of thyroid function in adults with down syndrome – 10-year follow-up study. *Journal of Intellectual Disability Research*, *51*, 312–317. doi:10.1111/j.1365-2788.2006.00879.x

Prasher, V.P., Huxley, A., Haque, M.S., & the Down Syndrome Ageing Study Group. (2002). A 24-week, double-blind, placebo-controlled trial of donepezil in patients with Down syndrome and Alzheimer's disease—pilot study. *International Journal of Geriatric Psychiatry*, *17*, 270–278. doi:10.1002/gps.587

Romeo, R., Knapp, M., Morrison, J., Melville, C., Allan, L., Finlayson, J., & Cooper, S.-A. (2009). Cost estimation of a health-check intervention for adults with intellectual disabilities in the UK. *Journal of Intellectual Disability Research*, *53*, 426–439. doi:10.1111/j.1365-2788.2009.01159.x

Schieve, L.A., Boulet, S.L., Boyle, C., Rasmussen, S.A., & Schendel, D. (2009). Health of children 3 to 17 years of age with Down syndrome in the 1997–2005 National Health Interview Survey. *Pediatrics*, *123*, e253–e260. doi:10.1542/peds.2008-1440

Sciberras, C., Vella, C., & Grech, V. (2004). The prevalence of coeliac disease in Down's syndrome in Malta. *Annals of Tropical Paediatrics*, *24*, 81–83. doi:10.1179/027249304225013268

Stephen, E., Dickson, J., Kindley, A.D., Scott, C.C., & Charleton, P.M. (2007). Surveillance of vision and ocular disorders in children with Down syndrome. *Developmental Medicine & Child Neurology*, *49*, 513–515. doi:10.1111/j.1469-8749.2007.00513.x

Swigonski, N.L., Kuhlenschmidt, H.L., Bull, M.J., Corkins, M.R., & Downs, S.M. (2006). Screening for celiac disease in asymptomatic children with Down syndrome: Cost-effectiveness of preventing lymphoma. *Pediatrics*, *118*, 594–602. doi:10.1542/peds.2005-2123

Unachak, K., Tanpaiboon, P., Pongprot, Y., Sittivangkul, R., Silvilairat, S., Dejkhamron, P., & Sudasna, J. (2008). Thyroid functions in children with Down's syndrome. *Journal of the Medical Association of Thailand*, *91*, 56–61.

Urv, T.K., Zigman, W.B., & Silverman, W. (2008). Maladaptive behaviors related to dementia status in adults with Down syndrome. *American Journal on Mental Retardation*, *113*, 73–86. doi:10.1352/0895-8017(2008)113[73:MBRTDS]2.0.CO;2

Van Cleve, S.N., & Cohen, W.I. (2006). Part I: Clinical practice guidelines for children with Down syndrome from birth to 12 years. *Journal of Pediatric Health Care*, *20*, 47–54.

van Schrojenstein Lantman-de Valk, H.M., Haveman, M.J., Maaskant, M.A., Kessels, A.G.H., Urlings, H.F.J., & Sturmans, F. (1994). The need for assessment of sensory functioning in ageing people with mental handicap. *Journal of Intellectual Disability Research*, *38*, 289–298.

van Trotsenburg, A.S.P., Heymans, H.S.A., Tijssen, J.G.P., de Vijlder, J.J.M., & Vulsma, T. (2006). Comorbidity, hospitalization, and medication use and their influence on mental and motor development of young infants with Down syndrome. *Pediatrics*, *118*, 1633–1639. doi:10.1542/peds.2006-1136

van Trotsenburg, A.S.P., Kempers, M.J.E., Endert, E., Tijssen, J.G.P., de Vijlder, J.J.M., & Vulsma, T. (2006). Trisomy 21 causes persistent congenital hypothyroidism presumably of thyroidal origin. *Thyroid*, *16*, 671–680.

Virji-Babul, N., Eichmann, A., Kisly, D., Down, J., & Haslam, R.H.A. (2007). Use of health care guidelines in patients with Down syndrome by family physicians across Canada. *Paediatrics & Child Health*, *12*, 179–183.

Weijerman, M.E., van Furth, A.M., Vonk Noordegraaf, A., van Wouwe, J.P., Broers, C.J.M., & Gemke, R.J.B.J. (2008). Prevalence, neonatal characteristics, and first-year mortality of Down syndrome: A national study. *The Journal of Pediatrics*, *152*, 15–19. doi:10.1016/j.jpeds.2007.09.045

Yam, W.K.-L., Tse, P.W.T., Yu, C.M., Chow, C.B., But, W.M., Li, K.Y., … Lau, J.T.F. (2008). Medical issues among children and teenagers with Down syndrome in Hong Kong. *Down Syndrome Research and Practice*, *12*, 138–140.

Zigman, W.B., & Lott, I.T. (2007). Alzheimer's disease in Down syndrome: Neurobiology and risk. *Mental Retardation and Developmental Disabilities Research Reviews*, *13*, 237–246. doi:10.1002/mrdd.20163