Primary Care Screening Tool to Support the Period Health Check in People with Down Syndrome

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ABSTRACT

Background: Down syndrome is the most common cause of intellectual disability in developed countries and is associated with several distinct physical features as well as health concerns such as congenital heart disease, gastrointestinal issues, sleep issues, thyroid problems, and vision and hearing problems. Primary care providers are expected to provide comprehensive care to their Down Syndrome patients despite limited training specific to caring for this population. There are guidelines available regarding regular screening for people with Down syndrome; however, not all are up to date nor easily found and integrated into a providers' electronic medical record. The objective of this project is to review the recommendations for screening in adults with Down syndrome and design a tool for family physicians to use during the Canadian Consensus Guideline recommended periodic health checks for their patients with Down syndrome to ensure their unique health needs are being met.

Methods: Eighteen articles were reviewed, and screening recommendations were synthesized into an evidence based tool for primary care providers to use during periodic health checks. The tool is a 5-page form that can be integrated into a physicians' electronic medical record and provided to the patient for tracking completed screening.

Results: Experts recommend annual screening for vision and hearing disorders, atlantoaxial instability, cardiac abnormalities, obesity, gastrointestinal disorders, dementia, and testicular and breast cancer. They also recommend regular assessment of dental hygiene, skin condition, thyroid function, autism, mental health concerns, sleep quality, and symptoms of menopause. Lastly, regular screening for diabetes, cardiovascular risk factors and cervical cancer are recommended as well as consideration of screening for osteoporosis.

Conclusion: This tool (Appendix I) was designed to help physicians facilitate regular health checks to increase health outcomes as recommended by the Canadian Consensus Guidelines. The tool

provides an easy check list for physicians to complete with their patients with Down syndrome with the goal of improving the long-term health outcomes of people with Down syndrome.

INTRODUCTION

Down Syndrome (DS) or Trisomy 21 is a genetic disorder that occurs naturally with a frequency of 1 per 781 births in Canada (Canadian Down Syndrome Society, 2024). It is due to a chromosomal arrangement resulting in an additional copy of chromosome 21. Down syndrome is the most common cause of intellectual disability in developed countries and is associated with a variety of distinct physical features as well as health concerns including congenital heart disease, gastrointestinal issues, sleep issues, thyroid problems, and vision and hearing problems (Canadian Down Syndrome Society, 2019; Moreau et al., 2021; Pikora et al., 2014; Toler, 2015). The life expectancy of an individual with Down syndrome was only 25 years in the 1980s, compared to an average of 60 years today thanks to medical interventions and treatments (Canadian Down Syndrome Society, 2024). As the life expectancy of individuals with DS increases, it is important to conduct regular relevant screening and intervention to optimize health outcomes.

Primary care providers are expected to have the knowledge to manage complex patients with the potential for multiple comorbidities in a single person on top of difficulty in locating and reporting symptoms as can be seen in people with Down syndrome. Despite this expectation, the likelihood that a primary provider has had limited training in managing individuals with Down syndrome or other intellectual or developmental disability is quite high. A tool that provides easy access to evidence-based information, structure to visits, guidance of the exam, as well as supports the management plan in the areas of screening, risk of health conditions, and suggested management would be an asset to any primary care provider who has patients with Down syndrome in their practice.

The objective of this clinical education tool is to create a resource for primary care providers in Nova Scotia to reference when managing adult patients with Down Syndrome to ensure their patients with Down Syndrome are receiving appropriate screening and intervention. This tool will support the primary care provider in conducting periodic health checks—healthcare visits with a focus on preventative counselling, immunizations, and known effective screening tests based on a patient's individual risks—for people with Down syndrome as recommended by the Canadian Consensus Guidelines. This tool can be used by physicians, nurse practitioners, nurses, physiotherapists, occupational therapist and any other health care professionals providing primary care. This topic was selected after working in a clinic specializing in providing care for adults with developmental and intellectual disabilities, recognizing my own lack of training in this area as a primary care provider and wanting to find a way to improve the care I provide to a patient population that I am not often exposed to, broadening my abilities as a skilled clinician. The tool I developed will allow for physicians, caregivers, and patients with Down syndrome to ensure the patient's healthcare needs are being addressed at regular intervals as determined by their primary care provider and adequate involvement of community-based resources such as optometrist, dentists and audiologists are being utilized.

BACKGROUND

The Canadian Consensus guidelines on primary care of adults with intellectual and developmental disabilities recommends routine periodic health assessments as a way to identify unmet needs and reduce mortality in individuals with intellectual and developmental disabilities such as those with Down syndrome (Sullivan et al., 2018). Health checks have been found to be the only intervention that leads to a significant increase in health actions, which then translates to an improvement in health outcomes at the primary care level (Ware et al., 2024). Regular health checks can serve as an educational tool and reminder for the person with DS as well as their

informal and formal support people, to monitor the individual's health and stay up to date on preventative healthcare (Ware et al., 2024). There are a variety of medical conditions that people with Down syndrome are more prone to developing that can impact their health long term.

HEENT

People with Down syndrome are more prone to developing certain ocular conditions including amblyopia, hyperopia, and astigmatism (Tishad et al., 2024), which can impact daily functioning. Similarly, ear and hearing disorders can impact communication and may occur earlier in people with DS due to accelerated aging experienced by this population (Belton et al., 2018; Canadian Down Syndrome Society, 2019). Hearing loss also further accelerates dementia, which people with Down syndrome are already at an increased risk of developing. In both adults and children with DS, tooth anomalies and gum disease, such as missing or malformed teeth, gingivitis and periodontitis are more common (Canadian Down Syndrome Society, 2019; Forster-Gibson & Berg, 2011; Moreau et al., 2021)

Endocrine Disorders

A number of thyroid diseases may be seen in the DS population including subclinical hypothyroidism, clinical hypothyroidism, hyperthyroidism, and autoimmune thyroiditis (Forster-Gibson & Berg, 2011). Approximately 1% of children with DS are born with congenital hypothyroidism and nearly 20% develop hypothyroidism after birth (Forster-Gibson & Berg, 2011). There is an increased prevalence of obesity in both children and adults with DS with most studies citing the rate of overweight and obesity as two to four times higher than that of the general population (Moreau et al., 2021; Tishad et al., 2024). The prevalence of diabetes in the DS population is higher than that of the general population (Bulova et al., 2020; Moreau et al., 2021). Both Type I diabetes and Type II diabetes are seen in the DS population and further study is required to determine whether Type I or Type II diabetes is particularly prevalent (Moreau et al., 2021)

Cardiac Disorders

Congenital heart defects (CHD) are well-known causes of morbidity and mortality in the Down syndrome population, affecting between 40% and 63.5% of individuals (Tishad et al., 2024). Given the prevalence of CHD, people with DS are at an increased risk of stroke. Although typical cardiovascular risk factors, such as hypertension, are not as prevalent in the adult DS population compared to risk factors such as obstructive sleep apnea (OSA) (Tishad et al., 2024).

Respiratory Disorders

Obstructive sleep apnea is extremely common among adults with DS with a nearly 100% prevalence, compared to a prevalence of only 5-8% in the general population (Moreau et al., 2021; Tishad et al., 2024). This is likely due to some of the distinct physical features associated with Down syndrome including macroglossia, adeno-tonsillar hypertrophy, midface hypoplasia, mandibular hypoplasia, hypotonia of the upper airway, small upper airway, obesity, and hypotonia (Dumortier & Bricout, 2020; Tishad et al., 2024).

Musculoskeletal Disorders

Atlantoaxial Instability (AAI) affects between 6.8% and 27% of the DS population (Tishad et al., 2024). It is caused by an extended distance between the rear surface of the frontal arcus of the C1 cervical vertebrae and anterior surface of the C2 cervical vertebra dens (Tishad et al., 2024).

Gastrointestinal Disorders

There are a number of GI issues that are seen in the Down syndrome population including constipation, gastroesophageal reflux disease, and celiac disease (Belton et al., 2018; Bulova et al., 2020; Tishad et al., 2024).

Neurocognitive Disorders

Autism spectrum disorder (ASD) has a prevalence of almost 2% in the general population, whereas studies conducted in people with DS the prevalence of ASD has been found to range from

1% to 19% (Brighenti et al., 2021). There is also an increased prevalence of Alzheimer's disease in people with Down syndrome secondary to the increased production of amyloid beta proteins coded for by the APP gene on chromosome 21 (Tishad et al., 2024). The mean age of diagnosis of Alzheimer's disease in the DS population is 55.8 years with symptoms usually beginning to manifest around age 40 (Bulova et al., 2020).

METHODS

A literature search was conducted using Ovid Medline and Psych Info to find articles pertaining to health screening in adults with Down syndrome. A filter was applied to exclude articles prior to 2014 to ensure information was up to date and that the number of articles generated was appropriate. Key words used included "Adult Development", "Physical Examination", "Health screening", "Health Outcomes", "Health Promotion", "Down's syndrome", "Down syndrome", "trisomy 21", "Adult", "Young Adult", and "Aged", which generated 34 articles from Ovid Medline and 93 articles from Psych info. The abstracts for all 127 articles were reviewed. Using exclusion criteria including "ID not specific to DS", "not related to DS" "Prenatal", "DS in childhood" and source types that were not journal articles, a total of 32 articles remained that were read in full. Of these articles, 11 were selected. An addition three articles were added based on supervisor recommendation, including a previous "Health Watch Table" from 2011. Two other articles were added following an external review of national and international Down syndrome organizations including an article by the Global Down Syndrome Foundation that included the Global Medical Care Guidelines for Adults with Down Syndrome Checklist (2020).

Final sources were reviewed and any recommendations pertaining to health screening and interventions for individuals with DS were documented. The screening recommendations based on the literature review were then synthesized into a table, which was sent to Dalhousie staff Carly Trumble, to be made into a form for Med Access, the electronic medical record (EMR) used by

Dalhousie family medicine. I worked with Carly Trumble to optimize the layout and appearance of the tool over email and ended with a 5-page document, 3 of which could be printed and provided to patients to track screening. In the case of screening guidelines that were not consistent across sources, priority was placed on the source that was most recent, and other recommendations were included underneath the main recommendation as an alternative screening option.

The decision was made to create a tool that could be easily integrated with the EMR, to avoid the bulkiness a macro containing the same information would cause, while keeping the tool readily accessible by primary care providers. A visual was also created to accompany some recommendations, which can be printed and provided to the patient. The intention here is to engage the patient in their preventative care with a tangible and interactive reminder. A copy of the tool can be printed for the patient's primary caregiver at relevant appointments as a physical reminder for screening that needs to be completed (See appendix for tool).

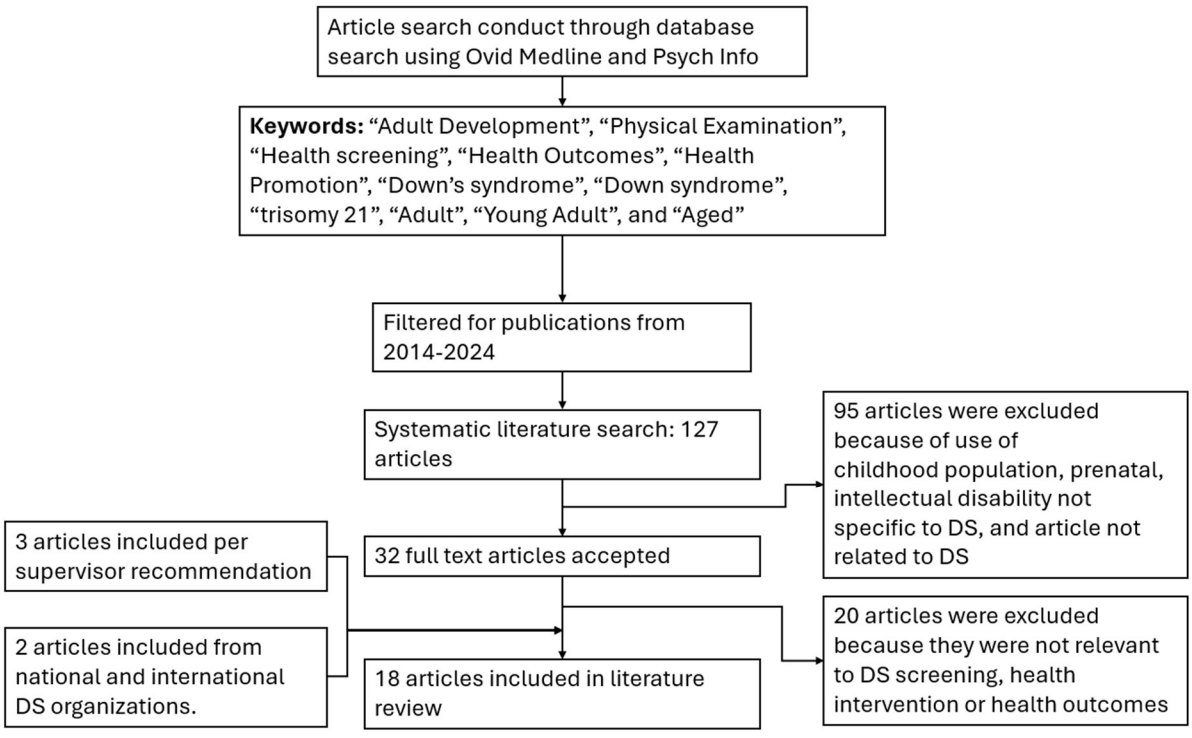

Figure 1. Literature review article selection process.

Previous Tools

The 2011 Health Watch Table for Down Syndrome by Forster-Gibson and Berg (Appendix II) is a two-column table with medical considerations on the left, grouped by system, and health recommendations on the right with a check box next to each recommendation. This was the original inspiration for the format of my tool; however, I wished to include more information such as date of completion for tests and results, which would be useful to primary providers when tracking screening. Therefore, I had to alter my layout and chose to organize my tool based on frequency of screening, grouping investigations that occur annually under one heading and grouping all other screening recommendations under another heading. This was chosen because it allowed the tool to be more succinct and appropriate for an EMR form.

The Global Down Syndrome Foundation created a similar guideline for screening in 2020 (Appendix III); however, they did not use a traditional table format and instead fit all recommendations on a single page using checkboxes and written instructions. As a result, the tool comes off as crowded and has very small text that may be difficult for the patient and/or provider to read.

RESULTS

A literature review was conducted to determine what health conditions adults with Down syndrome are at increased risk for developing and in which screening could be beneficial.

Eye Disease

In a 2014 study, Pikora et al. found that the most common conditions effecting their Down syndrome population were eye and vision concerns, which were seen in 72.6% of study participants. Belton et al. (2018) had similar results with over half of the older adults with DS in their study reporting an eye disorder. As a result, a recent article by Tishad et al (2024) recommends that vision be assessed annually or when new or worsening behaviour problems arise, as patients with

Down syndrome may have difficulty communicating when there is a problem or change in their health status. Alternatively, previous recommendations suggest vision testing be completed every 2 years in adults with down syndrome (Canadian Down Syndrome Society, 2019; Forster-Gibson & Berg, 2011).

Hearing Disease

Auditory evaluation, in office or by a specialist, should be completed annually as well as when new or worsening behavioural problems occur (Tishad et al., 2024). Once again, there is some debate regarding frequency of screening, and consideration can be made to screen every 2 years rather than yearly (Canadian Down Syndrome Society, 2019; Forster-Gibson & Berg, 2011).

Dental Disease

As mentioned in the introduction, tooth anomalies and gum disease, such as missing or malformed teeth, gingivitis and periodontitis are more common in people with down syndrome (Canadian Down Syndrome Society, 2019; Forster-Gibson & Berg, 2011; Moreau et al., 2021). As such, daily dental hygiene and dental evaluation every 6 months are recommended to help prevent gum disease (Forster-Gibson & Berg, 2011).

Thyroid Disease

The prevalence of thyroid disease continues to rise into adulthood as supported by Pikora et al. (2014), who observed that nearly a quarter of their young adult patients had thyroid disorders and Belton et al. (2018), who studied an older adult population found 43.8% of their subjects used thyroid medications, with 38.2% having a diagnosis of thyroid disorder. Due to the high prevalence of thyroid disorder in the DS population, comorbidity of presenting symptoms—such as fatigue, cold intolerance, obesity and constipation—and potential difficulty communicating, screening the DS population using TSH every 1-2 years starting at age 21 is recommended (Bulova et al., 2020; Tishad et al., 2024). A T4 can be completed at the same time or subsequently if TSH is abnormal

(Bulova et al., 2020). Consideration can be given to repeat T4 measurements every 6 months for 1 year in the setting of subclinical hypothyroidism (Forster-Gibson & Berg, 2011). Most sources agree that TSH should be checked annually in the DS population (Canadian Down Syndrome Society, 2019; Forster-Gibson & Berg, 2011).

Cardiovascular Disease

The neonatal echocardiogram has become a regular screening practice for people with Down syndrome, given the frequency of congenital heart disease; however, since it was only introduced in the mid-1980s, there are many adult DS patients who have undiagnosed CHD who may benefit from adulthood screening (Tishad et al., 2024). In addition, adults with Down syndrome are more prone to developing heart failure at an earlier age due to higher rates of valvular regurgitation and left ventricular thickening (Tishad et al., 2024). There is some variety when it comes to screening recommendations for cardiovascular disease in adults with DS. The consensus is that a regular cardiac examination should be conducted to assess for new abnormalities; however, the frequency of "regular" is not always defined. Forster-Gibson & Berg (2011) recommend an annual cardiac examination, with echocardiogram to confirm any new abnormal findings as well as regular monitoring for any people who had surgery in childhood. The Canadian Down Syndrome Society (2019) recommends an echogardiogram be compelted every 5 years in anyone with a history of cardiac surgery or ongoing heart issues as well as an annual blood pressure. Lastely, Tishad et al. (2024) recommends in adults with a normal childhood echocardiogram, a yearly cardiac examination is recommended and in those without a childhood screening echocardiogram, a one-time screening echocardiogram is recommended in addition to the yearly cardiac examination. Any abnormal findings should be further worked up with echocardiogram or referred to a cardiac specialist (Bulova et al., 2020; Forster-Gibson & Berg, 2011)

It is important to screen for and optimize modifiable risk factors for cardiovascular disease. Belton et al.(2018), found that high cholesterol was reported in 32.6% of their study sample, while obesity was present in 72.3%. In individuals with CHD, screening for atherosclerotic cardiovascular disease with a lipid panel and 10-year Atherosclerotic Cardiovascular Disease Risk Calculator, such as the Framingham, should begin at age 40 and occur every 5 years (Bulova et al., 2020; Tishad et al., 2024; Tsou et al., 2020).

Obesity

The increased prevalence of obesity in people with Down syndrome puts people with DS at risk for further health complications related to obesity such as Type 2 diabetes, gastrointestinal, and orthopaedic conditions (Pikora et al., 2014). The increased rate of obesity amongst people with DS may in part be explained by lifestyle, including a lack of physical activity, unhealthy dietary habits, and endocrine disorders (Moreau et al., 2021). People with DS have a reduced exercise capacity, not only as a result of lower peak of oxygen uptake, likely secondary to ventilatory dysfunction, but also muscle hypotonia, and ligamentous laxity that may impact body dynamics and balance (Moreau et al., 2021). In addition, people with DS may have leptin resistance secondary to increased leptin levels, which results in dysregulation of appetite and can contribute to obesity (Moreau et al., 2021). Therefore, all guidelines agree, it is important to evaluate weight or body mass index annually in the primary care setting (Bulova et al., 2020; Tishad et al., 2024; Tsou et al., 2020; Wilkinson et al., 2007). It is important to recognize that body mass index does not best predict both mortality and cardiovascular outcomes in middle-age and older-age adults and it may be beneficial to monitor waist-to-hip ratio instead as it has the best correlation between cardiovascular risk and all-cause mortality (Myint et al., 2014).

Obstructive Sleep Apnea

As mentioned in the introduction, obstructive sleep apnea is extremely common among adults with DS likely due to some of the distinct physical features associated with Down syndrome. Assessment of obstructive sleep apnea using polysomnography, should be conducted whenever an adult with DS presents with new-onset cognitive or behavioural changes, cardiovascular disease, or obesity (Tishad et al., 2024).

Diabetes

Immune system defects seen in DS likely contribute to the prevalence of Type I diabetes, whereas, the high prevalence of overweight and obesity likely impact the development of Type II diabetes (Moreau et al., 2021; Pikora et al., 2014). Regardless, regular screening is recommended in asymptomatic individuals with a hemoglobin A1c or fasting glucose every 3 years starting at age 30, in non-obese individuals, or every 2-3 years starting at age 21 in obese individuals (Bulova et al., 2020; Tsou et al., 2020).

Atlantoaxial Instability

Atlantoaxial instability can be asymptomatic or present with signs and symptoms of cervical myelopathy such as altered gait, urinary incontinence, hyperreflexia and muscle clonus (Tishad et al., 2024). Evaluation of signs and symptoms of cervical myelopathy should occur yearly with a targeted history and physical examination (Bulova et al., 2020; Tishad et al., 2024; Tsou et al., 2020). If a patient screens positive, a cervical spine radiograph should to ordered to confirm diagnosis (Bulova et al., 2020; Tishad et al., 2024; Tsou et al., 2020). Some sources recommend a lateral cervical spine X-ray, if not previously done, if an individual is participating in Special Olympics or is engaging in high-risk activities to prevent any potential damage (Canadian Down Syndrome Society, 2019; Forster-Gibson & Berg, 2011); however, no studies have been conducted that assessed whether screening asymptomatic adults with Down syndrome using cervical X-ray

impacts risk of spinal cord injury and Special Olympics organizers report no spinal cord injuries from over 50,000 individuals with DS who have participated in Special Olympics activities over 20 years (Bulova et al., 2020). Therefore, screening asymptomatic individuals prior to participation in Special Olympics and similar high-risk activities may be of low yield.

Osteoporosis

There are no clear guidelines for screening for osteoporosis in the DS population because, in general, adults with DS have a shorter life expectancy, lower bone mineral density, and greater risks for secondary causes of osteoporosis including exposure to antipsychotics, anti-epileptics, and proton pump inhibitors than the general population ((Belton et al., 2018; Bulova et al., 2020). Fredrigo et al. (2023) found men with DS had worse bone mineral density compared to women with DS, which they hypothesized may be due to the androgen deficiency seen in males with DS. It was also noted that prior to the age of 39, signs of osteoporosis were only present in the spine, while after age 39 changes were only seen in the femur (Fedrigo et al., 2023). This is likely due to the small sample sized of this study because it is unlikely any decreased bone mineral density in the spine would resolve or improve as an individual aged. Despite the increased prevalence of low bone mineral density the FRAX model would not be expected to accurately predict fracture risk in adults with Down syndrome, so a shared decision-making approach on this issue is recommended (Bulova et al., 2020; Tsou et al., 2020). The Canadian Down Syndrome Society, however, does recommend DEXA scans every 2 years for post menopausal women and suggests considering screening in men who have celiac disease or who are on medications which increase the risk of osteoporosis. In addition, anyone with DS who experiences a fragility fracture should be evaluated for secondary causes of osteoporosis, such as celiac disease, vitamin D deficiency, hypothyroidism, hyperparathyroidism and medications associated with adverse effects on bone health (Bulova et al., 2020; Tsou et al., 2020).

Gastrointestinal Complaints

There are a number of GI issues that are seen in the Down syndrome population including constipation, gastroesophageal reflux disease, and celiac disease (Belton et al., 2018; Bulova et al., 2020; Tishad et al., 2024). The Global Down Syndrome Foundation recommends screening for gastrointestinal and non-gastrointestinal signs and symptoms of celiac disease annually using targeted history and physical examination rather than serum testing. Special attention should be paid to GI symptoms such as constipation, loose stools, diarrhea, and abdominal cramping, in addition to behavioural changes, and rashes. Unfortunately, the GI symptoms are very general, non-specific, and common in the DS population. Serum Immunoglobulin A (IgA) and IgA anti-tissue transglutaminase (tTG) are commonly used to diagnose celiac disease in the general population; however, they are less clinically useful in patients with Down syndrome as routine cutoff values for diagnosis do not appear to correlate well with symptoms of celiac disease or intestinal biopsy results in people with DS (Bulova et al., 2020). As such, the Global Down Syndrome Foundation suggests screening based on symptoms as above, and if screening is positive, then move on to appropriate laboratory testing and consideration of biopsy as necessary. In some instances, they suggest a trial of a gluten-free diet at home; however, clinical decisions regarding need for small bowel biopsy and gluten avoidance are complex and should be discussed with the primary provider. Despite the unreliable nature of Celiac serum testing in the DS population, the Canadian Down Syndrome Society (2019) still recommends that people with DS be tested for celiac disease every three to five years.

Emotional/Psychiatric/Nervous disorders

In people with DS the prevalence of autism spectrum disorder (ASD) has been found to range from 1% to 19%, whereas in the general population the prevalence is around 2% (Brighenti et al., 2021). It is important to consider co-occurrence among neurodevelopmental disorders, especially

given that such co-occurrence may lead to an increase in behavioural problems including stereotypic, compulsive, self-injurious or aggressive behaviours (Brighenti et al., 2021). Brighenti et al. (2021) recommend screening for autism spectrum disorder routinely, and further investigations should a patient screen positive.

There is a well-known connection between Alzheimer's disease and Down syndrome with the mean age of diagnosis of Alzheimer's disease in the DS population being 55.8 years with symptoms usually beginning to manifest around age 40 (Bulova et al., 2020). However, Alzheimer's can be difficult to diagnose as signs and symptoms may or may not present the same way as in the general population. Furthermore, these people can be more complicated to assess if they have comorbid illnesses, challenges in communicating, increased severity of intellectual disability, and if reliable collateral information is unavailable (Tishad et al., 2024). As such, it is important to screen annually for changes from baseline function that could signal cognitive decline or dementia starting at age 40 (Bulova et al., 2020). Silverman et al. (2021) evaluated the National Task Group-Early Detection Screen for Dementia (NTG-EDSD)—a screening tool for dementia designed to be appropriate for individuals with varying ID severity— as a tool for screening for dementia in Down syndrome and found it was informative for distinguishing between cognitively stable adults and those with early clinical progression of AD confirming that it is a tool appropriate for screening for dementia in people with DS.

New or worsening behaviours, as outlined above, can be a sign of an underlying issue, for example, an undiagnosed neurocognitive disorder, cognitive decline, physical discomfort, or an underlying psychiatric concern, such as anxiety or depression. In addition, it is essential to recognize that not all people with DS will respond to the same issue with the same behaviours. Fonseca et al. (2014) found that after six subjects in their study lost their primary caregiver, they all exhibited behavioural changes and most subjects, even without loss of a caregiver, exhibited

behavioural changes over the course of their two-year study. People with DS are at increased risk of developing anxiety and depression due to factors such as smaller brain volume, suboptimal attachment style, neuropsychological deficits and depressive components of Alzheimer's disease (Tishad et al., 2024). If there is any concern of a mental health disorder, The Global Down Syndrome Foundation (2020) recommends referral to a clinician knowledgeable about both medical and mental health disorders as well as common behavioural characteristics of adults with Down syndrome. Alternatively, it is recommended to follow guidelines for diagnosis of mental health conditions in the Diagnostic and Statistical Manual of Mental Disorders 5th edition or the Diagnostic Manual-Intellectual Disability 2 (Bulova et al., 2020). In addition, they recommend annual reviewal of behavioural, functional, adaptive and psychological factors (Bulova et al., 2020).

Women's Health

As previously discussed, people with Down syndrome experience accelerated aging and as such, menopause may occur sooner, with the average age of diagnosis being 42 (Canadian Down Syndrome Society, 2019). There is often an assumption that people with DS are not sexually active or are infertile; however, this is a misconception. Fertility may be reduced in some women with Down syndrome, but many have regular menstrual cycles and are able to conceive and deliver children (Toler, 2015). Therefore, it is important to not overlook relevant health screening for people with breasts and uteruses. As in the general population, menopause is diagnosed clinically. The Canadian Down Syndrome Society recommends mammography and breast examination occur annually after the age of 40, or sooner if there is a significant family history of breast cancer. Lastly, routine pap smears should be conducted every 3 years in individuals aged 21 and older who are not sexually active, or annually for 2 years, then once every 3 years in those who are sexually active (Canadian Down Syndrome Society, 2019). However, it is important to factor in patient risk and

comfort and to consider individualized intervals for cervical cancer screening (Wilkinson et al., 2007).

In summary, there are not clear guidelines for screening individuals with Down syndrome and some screening recommendations vary more than others. It is important to use one's clinical judgement when determining appropriate screening for one's patients, considering a person's living situation, personal risk factors, and family history.

DISCUSSION

As is evident by the literature review, there are not universal recommendations for screening in people with Down syndrome. However, there are several conditions that the majority of specialists agree should be screened for including hearing and vision, dental health, cardiovascular health, obesity, thyroid disorders, dementia, gastrointestinal disorders, testicular cancer, and atlantoaxial instability. In addition, routine screening as in the general population should not be forgotten; however, the risk versus benefit must be considered given the special needs of this population and adjusting the frequency of screening may be appropriate. Regular health checks have been shown to increase health outcomes and by providing an easy check list for physicians to complete with their patients with Down syndrome the hope is that this will improve the long-term health outcomes of people with Down syndrome. This helps prevent both the patient and physician from feeling overwhelmed by the volume of screening as it can be built into an action plan and spread out over the period of a year on average.

The tool is a five-page checklist and accompanying visual checklist within Med Access. It includes the screening task, frequency of screening, completion status, date of completion, result if relevant, and additional notes if relevant. The tool is designed to be simple to use and is based on recommendations from experts within the field of caring for people with Down Syndrome. It allows a physician to visualize what screening is required and plan the spread of investigations over

multiple visits as appropriate. References are included within the tool and physicians can refer to the cited articles for further information if desired.

STRENGTH/LIMITATIONS

A strength of this tool is that it is succinct, built into the EMR, and easy to provide relevant information to the patient and their support system while also engaging the person with Down syndrome. A limitation is that some screening recommendations are not the most up-to-date, specifically regarding obesity, as research has come to light illustrating that Waist-to-hip ratio is a superior method of measurement compared to BMI. This is something to consider in the future as guidelines of obesity evolve and then adapting this tool accordingly. In addition, studies often do not include people with Down syndrome and more inclusive research is needed. This tool recommends the involvement of specialists knowledgeable in caring for individuals with Down syndrome; however, not all communities have access to such specialists and thus care may not be readily available. Finally, this tool has been created for Med Access and should it be widely disseminated, the tool would have to be altered to be compatible with other electronic medical records.

CONCLUSION

In conclusion, Down syndrome is the most common cause of intellectual disability in developed countries and many physicians lack appropriate training regarding screening for this unique population. This tool is designed to help physicians address this lack of knowledge in an easy and engaging way for the physician and patient.

Future considerations include producing a guide to aid physicians in conducting sensitive exams in this population where communication can be difficult. Alternatively, a guide for the patient's support system to have at home assessment conducted by individuals the patient is safe and comfortable with would be beneficial.

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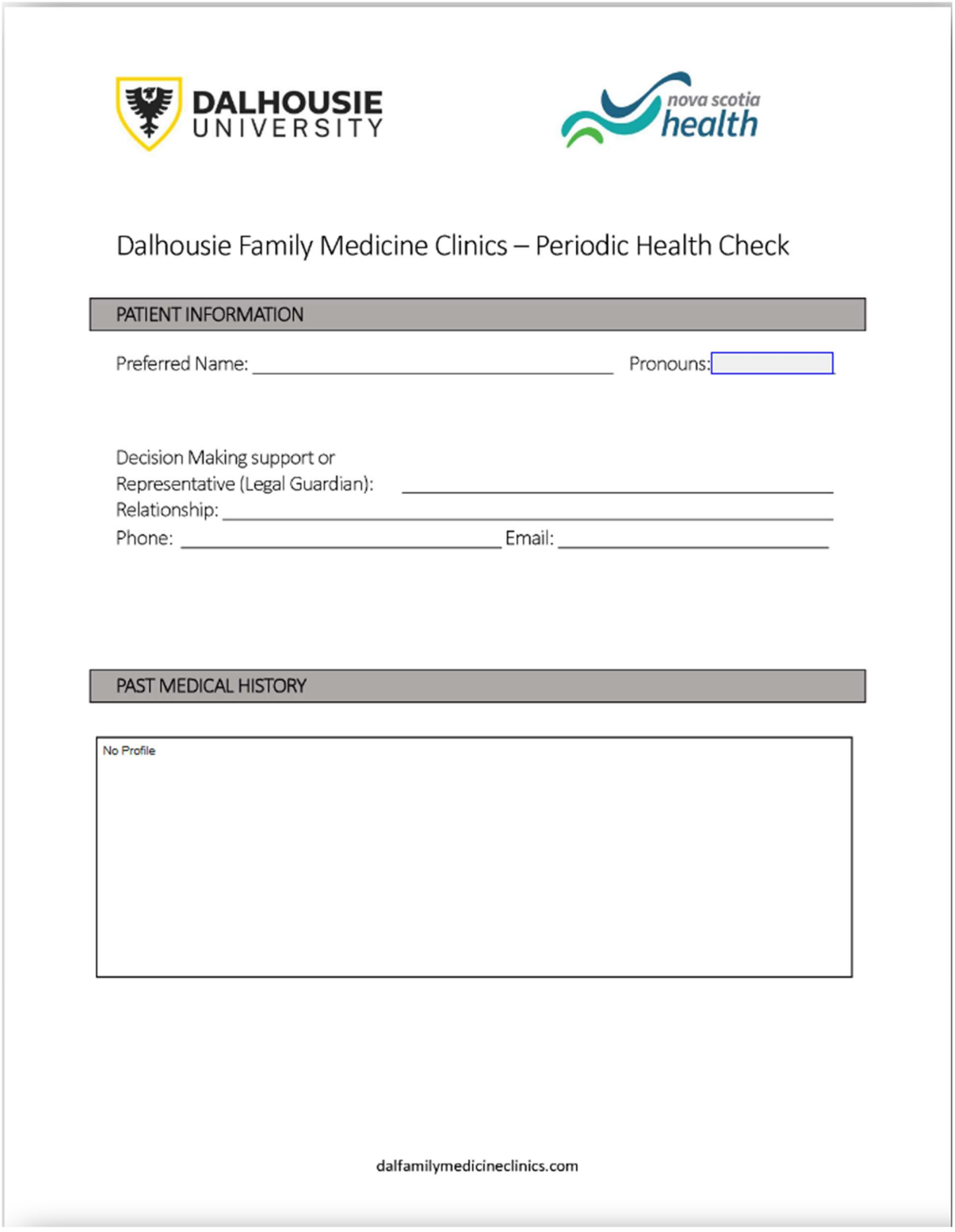
APPENDIX I. Down Syndrome Period Health Check Clinical Tool



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Dalhousie Family Medicine Clinics – Periodic Health Check

PATIENT INFORMATION

Preferred Name: _____ Pronouns: _____

Decision Making support or
Representative (Legal Guardian): _____
Relationship: _____
Phone: _____ Email: _____

PAST MEDICAL HISTORY

No Profile

dalfamilymedicineclinics.com

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TASKS

ANNUALLY	DATE COMPLETED	N/A
Vision Assessment[1,2] *Consider sooner if evidence of cognitive/behaviour change*		
Hearing Assessment[1,2] *Consider sooner if evidence of cognitive/behaviour change*		
Cervical Spine Assessment[1,3,4] *Movement, tenderness, pain, abnormal gait, urinary incontinence, hyperreflexia, muscle clonus* *Consider lateral cervical spine x-ray for dx of Atlantoaxial Instability (AAI) if sx as above*		
Cardiac Assessment +/- Echo[1,3–5] *One time echo if not completed in childhood OR* *Echo q5 years if ongoing cardiac issues or cardiac surgery history[5]*		
Weight/BMI[1–4]		
GI History[3] *Including symptoms of Celiac, constipation, reflux OR* *Celiac serum screening q3-5 years[5]*		
Testicular Exam[5,6]		
Breast Cancer Screening[5] *Mammography and breast exam annually after age 40; Sooner if significant family history of breast cancer OR Shared decision making*		
Dementia Screening (NTG-EDSD)[1,3] *Starting at age 40* *Screening tool: https://www.the-ntg.org/ntg-edsd*		
Review behavioural, functional, adaptive and psychosocial factors[3]		
TSH + Free T4[1,3,5,6] *Every 1-2 years starting at age 21* *OR every 6 months for 1 year if subclinical hypothyroid*	TSH: Free T4:	

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PREVENTATIVE CARE	DATE COMPLETED	N/A
Dental Hygiene[6] *Every 6 months* *Consider sooner if evidence of cognitive/behaviour change*		
Bone Mineral Density[3,4] *Shared decision making OR* *DEXA q2 years post menopause; consider in men if Celiac or increased risk of osteoporosis due to medications[5]*		
Skin Exam[2] *Routine care*		
Autism Screening (PDD-MRS/STA-HI)[7] *Routine Visits*		
Mental Health Screening[3] *As appropriate*		
Sleep Study[1] *Whenever evidence of new cognitive/behaviour change, CVD, or obesity*		
Symptoms of Menopause[5] *Average age for menopause in females with Downs syndrome is 42*		
Pap[5] *Pap and pelvic exam q3 once 21 if not sexually active; if sexually active annually for 2 years then q3 years OR Shared decision making*		
A1C or fasting plasma glucose[3,4] *Every 3 years starting at age 30* *OR every 2-3 years if obese starting at age 21*	HgbA1C: AC Glucose:	
Lipid Panel + Framingham[1,3,4] *Every 5 years starting at age 40*	Chol: HDL: LDL: Framingham Score:	

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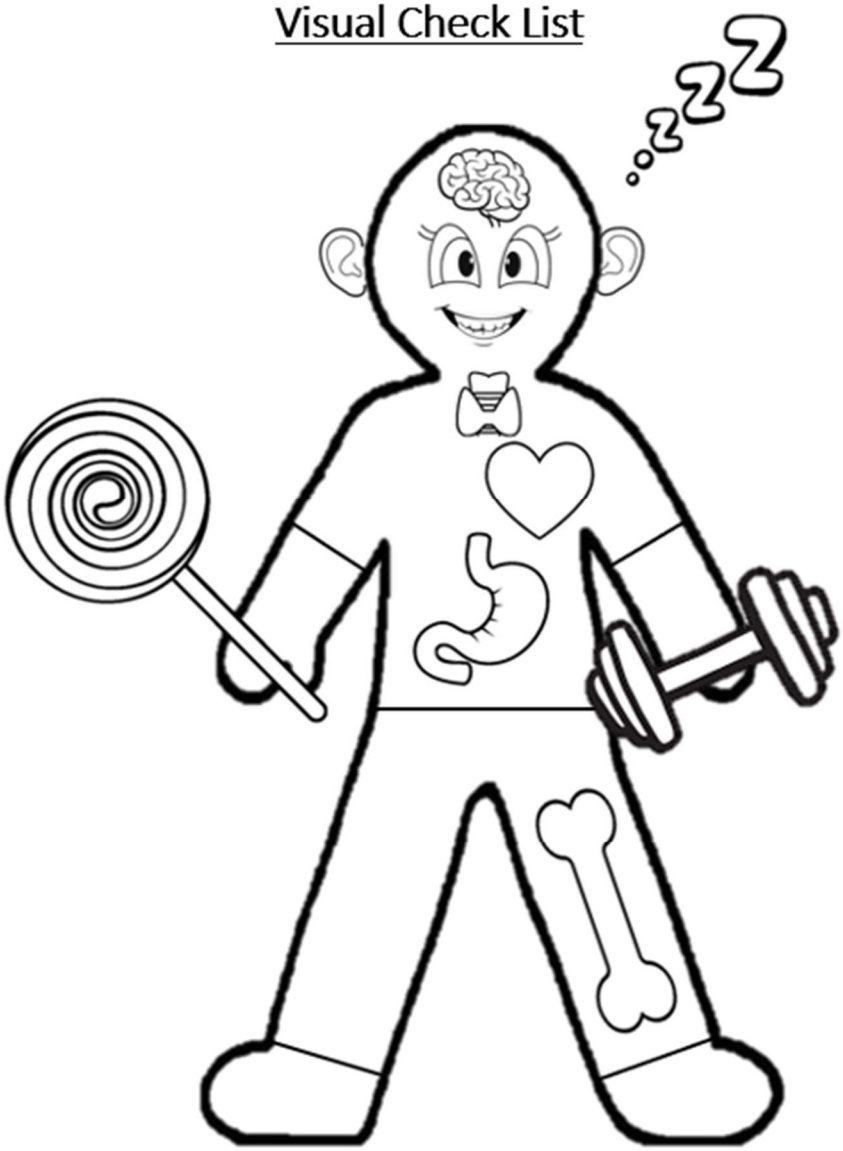
Visual Check List



- Dementia Screening
- Eye Exam
- Hearing Exam
- Dental Hygiene
- Thyroid Tests
- Heart Exam
- GI Assessment
- Diabetes Screen
- BMD
- Sleep Study
- Weight

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APPENDIX II. Health Watch Table

Health Watch Table — Down Syndrome

Forster-Gibson and Berg 2011

CONSIDERATIONS	RECOMMENDATIONS
1. HEENT (HEAD, EYES, EARS, NOSE, THROAT)	
Children and Adults: Vision: ~15% have cataracts; ~ 20% - 70% have significant refractive errors 5% - 15% of adults have keratoconus Hearing: 50% - 80% have a hearing deficit	☐ Neonatally: refer immediately to an ophthalmologist if the red reflex is absent or if strabismus, nystagmus or poor vision is identified. ☐ Arrange ophthalmological assessment: first by 6 months for all; then every 1-2 years, with special attention to cataracts, keratoconus, and refractive errors. ☐ During childhood: screen vision annually with history and exam; refer as needed. ☐ Arrange auditory brainstem response (ABR) measurement by 3 months if newborn screening has not been done or if results were suspicious. ☐ During childhood: screen hearing annually with history and exam; review risks for frequently occurring serious otitis media. ☐ Undertake auditory testing: first at 9 – 12 months, then every 6 months up to 3 years, annually until adulthood, then every two years.
2. DENTAL	
Children and Adults: Tooth anomalies are common Increased risk of periodontal disease in adults	☐ Undertake initial dental exam at 2 years, then every 6 months thereafter. Encourage proper dental hygiene. Refer to an orthodontist if needed. ☐ Undertake clinical exams every six months with referral, as appropriate.
3. CARDIOVASCULAR	
Children: 30% - 60% have congenital heart defects (CHD)	☐ Newborn screening: Obtain an echocardiogram and refer to a cardiologist, even in the absence of physical findings. ☐ In children and adolescents: review cardiovascular history and assess for physical signs with specialist referral, if indicated. • Refer for an echocardiogram if not previously done • Undertake SBE prophylaxis as indicated by findings
Adults: ~ 50% have cardiovascular concerns, commonly acquired mitral valve prolapse (MVP) and valvular regurgitation	☐ Ascertain a comprehensive cardiovascular history. ☐ Undertake an annual cardiac exam, with echocardiogram to confirm new abnormal findings and follow-up depending on the type of cardiovascular problem present or refer to an Adult Congenital Heart specialist or Disease clinic. ☐ Monitor regularly those that have had surgery in childhood. ☐ An echocardiogram is indicated to assess new abnormal physical findings or if unable to assess adequately by physical exam. Consider echocardiogram to establish baseline cardiac anatomy and function if not previously done or records are unavailable.[1]
4. RESPIRATORY	
Children and Adults: 50% - 80% have obstructive sleep apnea (OSA)	☐ Newborn: Refer to an ENT surgeon if recurring otitis media infections. ☐ Treat infections promptly and aggressively.
Adults: 50% - 80% have obstructive sleep apnea (OSA)	☐ Ascertain a detailed sleep history, with special attention to OSA symptoms. Refer to an ENT surgeon, including sleep study, if OSA is suspected. ☐ If aspiration pneumonia is suspected, investigate for possible swallowing disorder and gastro-esophageal reflux disease.

CONSIDERATIONS	RECOMMENDATIONS
5. GASTROINTESTINAL	
Children: ~ 50% have gastrointestinal (GI) tract anomalies including duodenal atresia, celiac disease, Hirschsprung disease, and imperforate anus	☐ Newborn: with vomiting or absent stools, check for GI tract blockage and refer to a gastroenterologist. ☐ Infants and children: anticipate constipation; treat with fluid/fibre/ laxative/stool softener/exercise/dietary change. ☐ From 2-3 years of age, screen for celiac disease. ☐ Establish good dietary and exercise habits to prevent or manage obesity.
Adults: ~ 95% are obese; ~ 7% have celiac disease	☐ Monitor for obesity. ☐ Screen for celiac disease, which may present in adulthood; screening tests used are the same as in the general population.[2] ☐ Test for Helicobacter Pylori and treat if positive, regardless of symptoms. ☐ Manage constipation proactively.
6. GENITOURINARY	
Children: Cryptorchidism is common	☐ Assess for hypogonadism, undescended testes, and possible testicular germ-cell tumors, or refer to a urologist, as appropriate.
Adults: Have increased risk of testicular cancer	☐ Assess annually by clinical exam, and refer to a urologist as appropriate.[3]
7. SEXUAL FUNCTION	
Adults: Fertility has been documented in women Fertility in males rarely reported	☐ Counsel regarding fertility possibility and the 50% [4] risk of Down syndrome in offspring.
8. MUSCULOSKELETAL (MSK)	
Children: ~15% have atlantoaxial instability (AAI)	☐ Arrange lateral cervical spine X-rays (flexed, neutral, and extended positions) between 3-5 years of age. ☐ Screen, as needed, prior to high risk activities (e.g., tumbling) and if participating in Special Olympics. ☐ Undertake an annual neurological exam for signs or symptoms of spinal cord compression. If present, refer urgently to a neurosurgeon and arrange an urgent MRI. ☐ Obtain a detailed MSK history with particular attention to possible joint subluxations/dislocations, scoliosis, and hip abnormalities.
Adults: Continued risk for spinal cord compression secondary to AAI Though data are limited, osteoporosis (associated with increased fractures risk) may be more common in older adults with Down syndrome than in similar aged individuals in the general population or with other developmental disabilities	☐ Undertake an annual neurological exam and assess for evidence of spinal cord compression. ☐ Arrange lateral cervical spine X-rays if not previously done, if presenting with signs and symptoms of AAI or if participating in Special Olympics. ☐ Take detailed history and attend to joint complaints, scoliosis, and hip abnormalities. ☐ If suspected, undertake bone mineral density (BMD) screening and refer to an appropriate specialist, if indicated. ☐ Encourage ambulation/mobility and weight reduction if obesity is present to decrease the risk of osteoarthritis.
9. NEUROLOGY	
Children: Epilepsy in up to 22%	☐ Take careful neurological history with particular attention to seizures (infantile spasms or tonic-clonic-type). ☐ Arrange an EEG and refer to a neurologist.

CONSIDERATIONS	RECOMMENDATIONS
Adults: Dementia is frequent and occurs earlier: 11%: 40 – 49 y, 77%: 60 – 69 y, Up to 75% with dementia have seizures with frequency increasing with age	☐ Obtain a neuropsychiatric history at every visit with particular attention to change in behaviour, loss of function/activities of daily living, and new onset seizures. ☐ If functional decline and/or signs/symptoms of dementia, use history, exam, and blood work to check for other conditions and treatable causes (e.g., hearing/vision deficits, obstructive sleep apnea, hypothyroidism, chronic pain, medication side effects, depression, menopause, low folic acid/vitamin B12). ☐ For possible seizures, arrange an EEG and refer to a neurologist.
10. DERMATOLOGICAL	
Children and Adults: Dry skin, atopic dermatitis, seborrheic dermatitis, chelitis, impetigo, and alopecia areata are more common than in general population	☐ Examine skin as part of routine care. ☐ Treat as per general population, with referral to dermatologist as needed.
11. BEHAVIOIURAL/MENTAL HEALTH	
Children: Self-talk is very common; autism spectrum disorder occurs in 5% - 10% of children with DS	☐ Review regularly with respect to behavioural concerns. ☐ Review for positive or negative signs suggestive of psychosis.
Adults: ~ 30% have a psychiatric disorder, including depression	☐ Review regularly with respect to behavioural concerns. ☐ Ascertain neuropsychiatric history at every visit, with particular attention to changes in behaviour, loss of function/activities of daily living, and new onset seizures.
12. ENDOCRINE	
Children: ~ 1% have congenital hypothyroidism; ~ 20% develop hypothyroidism after birth	☐ Review neonatal screening. ☐ Ascertain TSH and free T4 tests to confirm euthyroid status at 6 and 12 months, then annually. ☐ If signs of hyperthyroidism in adolescence, check for autoimmune thyroiditis.
Adults: 15% - 50% are hypothyroid Subclinical hypothyroidism, hyperthyroidism, and autoimmune thyroiditis are more common than in the general population	☐ For adults who are euthyroid, check TSH and free T4 levels at least once every 5 years [5] (some recommend annually).[6] ☐ If subclinical hypothyroidism (i.e., elevated TSH with normal free T4), follow free T4 every 6 months for one year [7] (some recommend treatment if thyroid antibodies are positive). ☐ Consider checking thyroid function whenever there are changes in mental status, behaviour or functional abilities.
13. HEMATOLOGICAL	
Children and Adults: Increased frequency of transient myeloproliferative disorder and leukemia No increased risk of leukemia in adults	☐ Neonates to 1 month olds: investigate for polycythemia and thromobocytopenia. ☐ Assess history periodically for symptoms of leukemia, with close attention to those with a history of transient myeloproliferative disorder.

WEBSITES THAT MAY BE HELPFUL FOR FAMILIES AND CAREGIVERS	
Canadian Down Syndrome Society	☐ www.cdss.ca/
Down Syndrome Education International [DownsEd]	☐ www.downsed.org/
Down Syndrome: Health Issues by Dr. Len Leshin	☐ www.ds-health.com/
Down Syndrome Medical Interest Group [DSMIG-UK]	☐ www.dsmig.org.uk/
National Down Syndrome Society [USA]	☐ www.ndss.org/

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APPENDIX D: GLOBAL MEDICAL CARE GUIDELINES FOR ADULTS WITH DOWN SYNDROME CHECKLIST (2020)

This checklist is intended to support the health of adults with Down syndrome directly or through their caregivers. We encourage this checklist to be shared with your medical professionals. Statements in blue represent our recommended, periodic health screenings/assessments that should begin at a specific age. Below each blue screening/assessment recommendation, there are blank boxes. Caregivers or individuals with Down syndrome can check off, date, or initial each blank box when the screening/assessment is completed. For screening/assessment recommendations with a time range (e.g. 1-2 years), the box size represents the longer possible time frame, such as 2 years versus 1. Statements in gray represent advisory recommendations that individuals with Down syndrome and caregivers should follow throughout adulthood.

Screening/Assessment | Advisory | Checkbox | No Recommendations

	21-29 Years	30-39 Years	40-49 Years	50-59 Years	60+ Years
Behavior	A review of behavioral, functional, adaptive, and psychosocial factors should be performed as part of an annual history that clinicians obtain from all adults with Down syndrome, their families, and caregivers. *(Boxes below represent 1 year increments)*				
	When concern for a mental health disorder in adults with Down syndrome is present medical professionals should: a) Evaluate for medical conditions that may present with psychiatric and behavioral symptoms and b) Refer to a clinician knowledgeable about the medical, mental health disorders, and common behavioral characteristics of adults with Down syndrome.				
	When concern for a mental health disorder in adults with Down syndrome is present, medical professionals should follow guidelines for diagnosis in the Diagnostic and Statistical Manual of Mental Disorders (DSM 5). The Diagnostic Manual-Intellectual Disability 2 (DM-ID-2) also may be used to adapt diagnostic criteria from the DSM-5.				
Dementia	Caution is needed when diagnosing age-related, Alzheimer's Type Dementia in adults with Down syndrome less than age 40.		Medical professionals should assess adults with Down syndrome and interview their primary caregivers about changes from baseline function annually beginning at age 40. Decline in the six domains as per the National Task Group – Early Detection Screen for Dementia (NTG-EDSD) should be used to identify early-stage age-related Alzheimer's-type dementia and/or a potentially reversible medical condition. *(Boxes below represent 1 year increments)*		
Diabetes		For asymptomatic adults with Down syndrome, screening for type 2 diabetes using HbA1c or fasting plasma glucose should be performed every 3 years beginning at age 30. *(Boxes below represent 3 year increments)*			
	For any adult with Down syndrome and comorbid obesity, screening for type 2 diabetes using HbA1c or fasting plasma glucose should be performed every 2-3 years beginning at age 21. *(Boxes below represent 3 year increments)*				
Cardiac			For adults with Down syndrome without a history of atherosclerotic cardiovascular disease, the appropriateness of statin therapy should be assessed every 5 years starting at age 40 and using a 10-year risk calculator as recommended for adults without Down syndrome by the U.S. Preventive Services Task Force. *(Boxes below represent 5 year increments)*		
	For adults with Down syndrome, risk factors for stroke should be managed as specified by the American Heart Association/American Stroke Association's Guidelines for the Primary Prevention of Stroke.				
	In adults with Down syndrome with a history of congenital heart disease, given the elevated risk of cardioembolic stroke, a periodic cardiac evaluation and a corresponding monitoring plan should be reviewed by a cardiologist.				
Obesity	Healthy diet, regular exercise, and calorie management should be followed by all adults with Down syndrome as part of a comprehensive approach to weight management, appetite control, and enhancement of quality of life.				
	Monitoring for weight change and obesity should be performed annually by calculating Body Mass Index in adults with Down syndrome. The U.S. Preventive Services Task Force Behavioral Weight Loss Interventions to Prevent Obesity-Related Morbidity and Mortality in Adults should be followed. *(Boxes below represent 1 year increments)*				
Atlantoaxial Instability	In adults with Down syndrome, routine cervical spine x-rays should not be used to screen for risk of spinal cord injury in asymptomatic individuals.				
	Annual screening for adults with Down syndrome should be based on a review of signs and symptoms of cervical myelopathy using targeted history and physical exam. *(Boxes below represent 1 year increments)*				
Osteoporosis	For primary prevention of osteoporotic fractures in adults with Down syndrome, there is insufficient evidence to recommend for or against applying established osteoporosis screening guidelines, including fracture risk estimation; thus, good clinical practice would support a shared decision-making approach to this issue would support a shared decision-making approach to this issue.				
	All adults with Down syndrome who sustain a fragility fracture should be evaluated for secondary causes of osteoporosis, including screening for hyperthyroidism, celiac disease, vitamin D deficiency, hyperparathyroidism and medications associated with adverse effects on bone health.				
Thyroid	Screening adults with Down syndrome for hypothyroidism should be performed every 1-2 years using a serum thyroid-stimulating hormone (TSH) test beginning at age 21. *(Boxes below represent 2 year increments)*				
Celiac Disease	Adults with Down syndrome should receive an annual assessment for gastrointestinal and non-gastrointestinal signs and symptoms of celiac disease using targeted history, physical examination and clinical judgement of good practice. *(Boxes below represent 1 year increments)*				

This checklist is not intended to be diagnostic. Presentation of medical and mental health conditions for people with Down syndrome may be atypical. Similar signs and symptoms may be a consequence of multiple reasons, including different disease processes. Thus, the patient evaluation should include considerations of additional causes for any detected sign or symptom. The development of new and/or changes in signs or symptoms should prompt a comprehensive evaluation with your clinician.