

Behaviour changes in an adult with Down syndrome

Cynthia J. Forster-Gibson MD PhD

Adults with Down syndrome are at increased risk of depression, hypothyroidism, and Alzheimer dementia (AD) as they age, all of which can cause behaviour changes. Basal ganglia calcification (BGC), similar to that seen in the rare condition of primary familial brain calcification (PFBC), is not uncommon in people who have Down syndrome and might be present throughout the life span. In an adult with Down syndrome who has behaviour changes, it is important to fully assess medical concerns common to adults with this condition that could affect behaviour, and to avoid premature investigation of rare conditions.

Challenges in the primary care of aging adults with developmental disability can include limited information on differences in the aging process; complex health needs; causes of developmental delay; behaviours that challenge; and communication problems. These topics are discussed in the 2 special issues of *Canadian Family Physician* on the primary care of adults with intellectual and developmental disabilities (IDD),^{1,2} particularly in "Circles of care for people with intellectual and developmental disabilities."³ Understanding the natural history of a condition that causes developmental differences is important, particularly when there is an increased probability of specific medical concerns. Health Watch Tables have been developed for several genetic and nongenetic causes of developmental delay⁴ and are useful for family members, caregivers, adults with developmental delay, and health care providers to understand and integrate the health care needs of adult patients with these conditions.

Case

Consent and assent for this report was obtained from Mr L. and his family. Mr L., a 43-year-old man who has Down syndrome, had a 2-year history of behaviour changes, paranoia, and auditory hallucinations, as well as losses in his social, work, and daily living skills. His caregivers had noticed increased fatigue and a change in his gait and speech. He had previously had a full-time job, maintained friendships, and lived independently with some support, but was now working less, had lost friends, and was having trouble living independently.

A computed tomography (CT) scan of his brain was arranged. The report described the presence of BGC and raised the possibility of PFBC. Mr L. was referred to a genetics service to investigate the possibility of PFBC, given the CT findings and what was thought to be an atypical presentation of dementia. He was seen in the genetics service, along with his mother and support worker.

Other than having been treated for a tumour at age 30, Mr L. had otherwise been in good health. At age 72, Mr L.'s father had had 2 strokes. A maternal uncle and aunt reportedly had had late-onset AD, and a paternal grandparent had had multi-infarct dementia. There was no known family history of PFBC.

In addition to the psychiatric services he had received, Mr L.'s family physician had referred him to a dual diagnosis program.

Mr L.'s vision and hearing had not been assessed lately. Recently measured glucose, urea, creatinine, electrolyte, alanine aminotransferase, alkaline phosphatase, lipase, and magnesium levels and toxicology screening findings were normal.

On examination, Mr L. had a wide-based, shuffling gait; mild spasticity in the lower extremities; and a mild decrease in strength in the legs. There were no cerebellar signs and no tremor or choreoathetosis.

He was taking olanzapine daily, which his caregivers believed was quite sedating.

Editor's key points

- ▶ The diagnostic assessment of adults with Down syndrome for Alzheimer dementia is similar to accepted practice in the general population. Caveats include limitations on the usefulness of neuropsychological assessment tools and neuroimaging, and the common occurrence of hypothyroidism and depression.
- ▶ Knowledge of common health problems associated with a specific condition causing intellectual and developmental disability can improve the approach to primary health care and assessment of changes in behaviour.
- ▶ Health Watch Tables and other tools developed specifically for adults with intellectual and developmental disability can guide their primary health care.

Points de repère du rédacteur

- ▶ L'évaluation diagnostique de la démence d'Alzheimer chez des adultes atteints du syndrome de Down est semblable aux pratiques acceptées pour la population en général. Au nombre des différences à signaler figurent l'utilité limitée des outils d'évaluation neuropsychologique et de la neuroimagerie, de même que la présence courante d'hypothyroïdisme et de dépression.
- ▶ La connaissance des problèmes de santé courants associés aux facteurs particuliers qui causent la déficience intellectuelle et développementale peut améliorer la façon de prodiguer des soins primaires et d'évaluer les changements comportementaux.
- ▶ Des tableaux de surveillance de la santé comme les Health Watch Tables et d'autres outils précisément élaborés pour les adultes ayant une déficience intellectuelle et développementale peuvent orienter la prestation de leurs soins primaires.

Discussion

Primary familial brain calcification is a progressive neuropsychiatric and movement disorder with bilateral BGC. Psychiatric symptoms range from mild memory problems to dementia and psychosis.⁵ Migraines are frequent and seizures might occur. Primary familial brain calcification might be familial (typically autosomal dominant) or sporadic. The prevalence is unknown, although more than 100 kindreds and sporadic cases have been reported.⁵

Similar patterns of BGC can occur secondary to infection and other nongenetic and genetic causes. Ramos et al⁵ offer a comprehensive discussion. Basal ganglia calcification occurs incidentally in the general population, particularly the elderly, appearing in 0.3% to 1.5% of CT scans.⁵ In persons with Down syndrome, it has been found on histopathology in 45% to 87% of patients^{6,7} and on CT scan in 10.7% to 27% of patients^{8,9} and has been found at all ages.⁶ There is no clear correlation between the presence of BGC and neuropsychiatric symptoms,¹⁰ including AD.

In persons with Down syndrome, pathology studies of the brain have shown the presence of neurofibrillary tangles and plaques typical of AD in 7.5% of teens, 80% of adults in their 30s, and almost all adults older than 40 years of age,¹¹ although the condition might not be symptomatic. The prevalence of diagnosed AD in adults with Down syndrome has been estimated to be 9.4% for those aged 40 to 49, increasing to 54.5% for those aged 60 to 69.¹²

Given the increased incidence of depression, hypothyroidism, AD, and hearing and vision impairments in persons with Down syndrome (described in the Down syndrome Health Watch Table),⁴ it was thought that 1 or more of these factors was more likely to be occurring in Mr L. than PFBC and all should be assessed. While it is not known why BGC occurs in, or how it affects the health of individuals with, Down syndrome, BGC is unlikely to indicate a separate rare condition, such as PFBC.

Differentiating between depression and early-stage AD can be very difficult, and depression can be a sign of early AD. Progressive changes that might suggest early-stage AD include loss of language, behaviour changes (including social withdrawal), loss of daily living skills, gait disorder, and, in some, psychosis (including hallucinations and delusions), seizures, and dysphagia. Investigation for AD in an adult with Down syndrome is similar to that for persons without Down syndrome,¹² although neuroimaging and psychological testing have specific limitations.^{12,13} Function should be tracked over time, and this could include use of a modified standardized tool.¹³⁻¹⁵ Vision and hearing impairments might change perception, contributing to symptoms suggestive of dementia, and should be assessed. Recommended blood tests include complete blood count; liver, renal, and thyroid function tests; and measurement of folate, vitamin B12, blood glucose, and electrolyte levels, and lipid profile.^{12,16} If clinically indicated, syphilis and HIV status should be determined.¹⁶ Diagnosis is primarily based on clinical assessment.

Conclusion

Assessment of behavioural changes in adults with Down syndrome should begin with assessment of medical concerns common to Down syndrome. Alzheimer dementia is seen earlier in persons with Down syndrome than in the general population and should be investigated in a similar fashion, with consideration of using tools for dementia assessment developed for individuals with IDD. The behavioural changes seen for Mr L. were not atypical for AD but could also suggest, or be exacerbated by, depression or other medical concerns, and he benefited from investigation for those and appropriate treatment. It was determined that PFBC would be very unlikely to be present.

When Mr L. was seen, the opportunity arose to discuss the Developmental Disabilities Primary Care Initiative's tools for the primary care of people with IDD.¹⁷ Mr L.'s doctor had retired and he had recently found a new doctor. The family members were very happy to be able to take information from the tools to their new doctor for further discussion and help in Mr L.'s future health care. 🌟

Dr Forster-Gibson is a general practitioner practising in clinical genetics, Physician Lead in the Genetics Program at Trillium Health Partners in Mississauga, Ont, and Assistant Professor in the Department of Laboratory Medicine and Pathobiology at the University of Toronto in Ontario.

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Competing interests

None declared

Correspondence

Dr Cynthia J. Forster-Gibson; e-mail Cynthia.Forster@thp.ca

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