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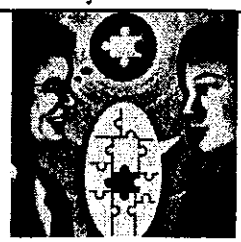
Abstract:

This article can be useful both for general practitioners and families and carers of adults with intellectual disability. Nicholas Lennox -Director of the QLD Centre for Intellectual and Developmental Disability, and Gillian Eastgate - Senior Lecturer at the same centre of The University of QLD, emphasise the important role that carers and people who are close to the person with intellectual disability can play to improve the quality of life of the person with disability. Carers and family can collaborate with the GP by providing key-information about habits, family history and other details that the practitioner could not gather during the consultation but may be essential to improve the health of the patient. The article includes information about the conditions and activities commonly and easily missed that may affect the health of the person with intellectual disability.



Adults with intellectual disability and the GP

BACKGROUND General practitioners are the health professionals most commonly consulted by people with intellectual disability. This group of patients can provide unique but not insurmountable challenges to the GP.

OBJECTIVE This article outlines the management of this group of patients, and provides strategies for treatment using a case vignette.

DISCUSSION While people with intellectual disability frequently have unidentified and/or sub-optimally managed conditions, improvement in their health care can be made through a variety of strategies. These include maximising communication and cooperation with all those involved and ensuring adequate information from support staff. Regular health assessments, which specifically target the commonly associated comorbidity and health screening activities, are also a useful approach. Through collaboration and proactive health care, the quality of life of patients with an intellectual disability can be substantially improved.

Case history – Reg

Reg shuffled into my consulting room and flopped onto the chair. Three jeans-clad carers filed in behind him, chatting among themselves as if on a shopping trip. Reg looked across at me, body tense, eyes pleading.

'How can I help you?' I asked Reg. He waved an arm and let out a loud moan. I looked across to his carers – they shrugged. Reg's main carer was on holiday. Only she was able to understand him. They had no idea. What they did know was that Reg had always been difficult to handle and at times became aggressive; over the past 3 months he had become increasingly agitated and disturbed. He frequently hit himself and anyone else within reach.

My heart sank. New patient, symptoms unknown, communication problems and a 15 minute appointment. I was already running an hour behind. This was going to take forever – and would it change anything for Reg?

Reg had lived in an institution from childhood until a few years ago. When the institution closed, he was moved into supported community accommodation and was now living in a rented house with three other men, each with an intellectual disability.

Because of his aggressive behaviour he had been moved from house to house, and none of his carers had known him for more than 9 months. They knew little about his past medical history. They had brought him to see me because they had decided they did not like the last general practitioner.

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This didn't give me much to go on. Examining Reg was similarly difficult. He did not like being touched, and especially disliked the abdominal examination. There were no findings of note. A plain abdominal X-ray revealed the cause of his distress.

He had a severely overloaded, distended colon, the result of years of severe constipation. It wasn't hard to guess the cause: several types of major tranquillisers, poor fluid intake and limited diet.

Reg was admitted to hospital for a manual extraction and revision of his medications. He required ongoing close monitoring of his bowels by his care staff. His behaviour improved dramatically and his carers reported that he was 'a different person'.

How could this situation have been prevented?

Like many adults with intellectual disability, Reg has been disadvantaged by instability in his living situation, and by lack of coordinated health care. He is at risk of undetected and undiagnosed physical and mental health conditions. Even when recognised, health problems may be poorly managed. Adults with intellectual disability on average have 5.2 conditions per person; half of these go unrecognised or are poorly managed.[1]

To improve coordination of his care, Reg needs to develop a relationship with one general practitioner, and to see that GP each consultation if possible. Comprehensive records should be available to other GPs in the practice for when his regular GP is unavailable. The GP needs to commit to providing ongoing and preventive care. He or she also needs to be aware of the atypical ways illness may present in this group of patients.

Reg's carers need to obtain as much information as possible, and to bring this information to each consultation. Reg should visit his GP with someone who knows him well. Carers need to be aware that the GP is not paid for fact finding work outside the consultation.

Once Reg's presenting problem has been addressed (and this may take more than one consultation), he should be assessed for other unrecognised health problems (*Table 1*).[2] A comprehensive health review (possibly utilising a Care Plan under the Medicare Extended Primary Care items) may save time in the longer term. An effective review requires a detailed past history – including developmental history, medications, health screening and preventive activities – and detailed information about any specific medical conditions including seizure frequency and type for patients with epilepsy, bowel history or behaviour history. Family history is important, especially when the disability aetiology is not known; GPs play an important role in detecting inherited conditions such as fragile X syndrome.

Reg's social environment should be considered. Multiple moves, staff changes, and possible physical and sexual abuse (from staff or co-residents) are all stressful events that commonly occur in the lives of people with intellectual disability. It is also important to assess how well Reg is functioning in both his current work and accommodation environments, and whether he could gain greater independence with appropriate assistance.

Ongoing management

It is important to ascertain the cause of the patient's disability, as some syndromes are associated with specific conditions (eg. hypothyroidism and heart anomalies in Down syndrome).[3] People with obvious dysmorphic features, more severe intellectual disability or other associated conditions are more likely to have a definable cause than those with milder disability and no dysmorphism. Where the cause of the disability is unknown, referral for genetic review and assessment by a metabolic physician is recommended.[4] If the cause is known, associated conditions can be addressed (see *Resources*).

Strategies to improve Reg's living environment or to teach him new skills – especially in communication – may improve his life considerably.

Management tips

- 'Listen' with all your senses
- Expect accurate information from caregivers. This is part of their responsibility. If reliable information is not forthcoming, contact senior management with your concerns
- Always fully examine the patient. It is easy to miss something in a patient who has problems communicating
- Sudden behaviour changes are almost always a communication of need or distress

Table 2. Syndrome specific list for GPs

	Cerebral palsy 1:500	Down syndrome 1:800	Fragile X 1:4000	Prader-Willi 1:10 000-25 000	Phenylketonuria 1:10 000-1:20000
Audiovisual	Visual impairment Hearing impairment	Eye pathology and visual impairment (multifactorial), cataracts Hearing impairment (multifactorial) Both very common (annual assessments recommended)	Visual impairment (multifactorial) Hearing impairment Recurrent ear infections	Strabismus Myopia	
Endocrine	Osteoporosis	Hypothyroidism (annual TFT recommended) Osteoporosis		NIDDM (secondary to obesity) Hypogonadism Delayed puberty	
Psychiatric/ psychological	Depression Variable intellectual capacity	Depression Alzheimer type dementia (20 years earlier than general population)	Attention deficit/ hyperactivity Variable intellectual capacity Disabled in social functioning Poor eye contact Hand flapping	Hyperphagia needs active monitoring to prevent morbid obesity, associated morbidities and death Impulse control difficulties Self injury	Variable intellectual capacity Phobic anxiety Disabled in social functioning Behaviour difficulties aggression, self injury
Central nervous system	Epilepsy	Epilepsy Usually clonic/tonic	Epilepsy Usually clonic/tonic, complex partial	Hypotonia as neonate Small hands and feet Short stature Almond shaped eyes	Epilepsy Hyperactivity Tremor and pyramidal tract signs Extrapyramidal syndromes
Cardiovascular		Congenital heart defects (~ 50%)	Aortic dilatation, mitral valve prolapse		
Musculo-skeletal and skin	Orthopaedic problems Neuromuscular problems Pressure areas skin	Atlantoaxial instability <1% Skin disorders, alopecia, eczema	Connective tissue dysplasia Scoliosis Congenital hip dislocation	Scoliosis, kyphosis Hypotonia Skin picking	
Other	Genito-urinary problems Incontinence Constipation Dental problems Recurrent aspiration Oesophagitis, gastroesophageal Reflux +/- bleeding, anaemia Swallowing, eating difficulties	Blood dyscrasias Childhood leukaemia Sleep apnoea Obesity Susceptibility to infections Coeliac disease	Herniae Abnormalities of speech and language Characteristics more distinctive in males may not appear until late childhood Macro-orchidism, large head, prominent ears, long face	Infantile failure to thrive, then hyperphagia and severe obesity High tolerance to pain Decreased ability to vomit Poor body temperature control Sleep apnoea Osteoporosis Undescended testes Dental abnormalities	Eczema Improvement has been reported in previously untreated adults after introduction of a phenylalanine restricted diet
Inheritance	90% cause by antenatal problem ~10% anoxia at birth	95% nondisjunction at miosis leading to trisomy chromosome 21; 4% translocation of C21 or rarely parental mosaicism C21	X-linked	Chromosome 15 – parental deletion 70%, uniparental disomy 25% and imprinting defect 5%	Autosomal recessive

Table 2. Syndrome specific list for GPs

	Angelman syndrome <1:10 000 <20 000	Williams <1:20 000	Rett 1:14 000 females	Noonan <1:10 000	Tuberous sclerosis 1:6 000-17 000	Neurofibromatosis 1:3000
Audiovisual	Glaucoma	Hyperacusis Strabismus	Refractory errors	Strabismus refractive errors Vision/hearing impairments	Retinal tumours Eye rhabdomyomatas	Hearing impairment (glioma affecting auditory nerve)
Endocrine						Various endocrine abnormalities
Psychiatric/ psychological	Easily excitable Hyperactive	Variable intellectual capacity Attention deficit problems in childhood	Severe intellectual disability	Mild intellectual disability	Variable intellectual capacity Behavioural difficulties Sleep problems	Variable intellectual capacity
Central nervous system	Severe developmental delay apparent by 1 year Microcephaly Epilepsy	Perceptual and motor function reduced	Epilepsy Vasomotor instability	Epilepsy	Cerebral astrocytomas Epilepsy	Variable clinical phenomena depending on site of the tumours Epilepsy
Cardiovascular		Cardiac abnormalities Hypertension, CVAs Chronic hemiparesis	Prolonged QT interval	Pulmonary valvular stenosis ASD, VSD, PDA	Rhabdomyomatas Hypertension	
Musculo-skeletal	Joint contractures and scoliosis (in adults) Truncal hypotonia Limb hypertonia	Joint contractures Scoliosis Hypotonia	Osteopenia Fractures Scoliosis	Scoliosis Talipes equinovarus Pectus carinatum/ excavatum	Bone rhabdomyomata	Skeletal abnormalities esp. kyphoscoliosis
Other	Speech impairment Movement and balance disorder Characteristic EEG changes	Renal abnormalities	Hyperventilation Apnoea Reflux Feeding difficulties Growth failure	Abnormal clotting factors, platelet dysfunction Undescended testes, deficient spermatogenesis Lymphoedenoma Hepatosplenomegaly Cubitus valgus, hand abnormalities	Kidney and lung hamartomata Polycystic kidneys Liver rhabdomyomata Dental abnormalities Skin lesions	Variable clinical phenomena depending on location of the neurofibroma Tumours are susceptible to malignant change Other varieties of tumours may be associated
Inheritance	Variety of genetic mechanisms on chromosome 15	Microdeletion on chromosome 7	Mainly sporadic	Autosomal dominant may be sporadic	Autosomal dominant	Autosomal dominant

Table 1. Conditions and activities commonly and easily missed

Psychiatric disorders
- Depression
- Schizophrenia
- Bipolar affective disorder
- Anxiety disorders
- Post-traumatic stress disorder

Gastrointestinal disorders
- Constipation/atonic bowel
- Bowel obstruction
- Reflux oesophagitis
- H. Pylori infection

Undescended testis/hypogonadism

Unrecognised pain or infection
- Dental pathology
- Chest infection
- Urinary tract infection

Medication issues
- Overuse of tranquillisers
- Unrecognised side effects of medication

Epilepsy management
- Inadequate review of anticonvulsant medication
- Failure to consider medication interactions and toxicity

Sensory impairment
- Hearing impairment
- Visual impairment
- Ear and eye pathology

Health maintenance activities
- Immunisation
- Screening for infectious conditions including hepatitis B
- Nutritional assessment (exclude malnutrition and obesity)
- Breast checks and Pap tests
- Blood pressure and skin checks
- Screening for osteoporosis and vitamin D deficiency where appropriate
- Physical activity assessment

- Acknowledge the patient's right to choose (eg. consent to examination or tests) even if he or she is unable to communicate verbally
- Patients with disabilities should be managed to the same standard as anyone else. Consider whether you would manage this problem the same way in a patient without a disability. If not, ask yourself if the patient is being devalued
- Insist on follow up appointments
- Consider common missed conditions (*Table 1*)
- Minimise polypharmacy
- Consider the psychosocial as well as biological aspects of care.

Behavioural issues

Behaviour change is a common reason for seeking medical care. Usually the person is trying to communicate that they are distressed. Causes for distress include illness, environmental changes, and/or psychological problems.[5] Physical and mental health problems need to be addressed and, if these are found not to be the reason for the change, a functional assessment of the patient's behaviour should be undertaken. Trained psychologists usually perform these assessments, however, it is not uncommon for such an assessment to be unavailable or difficult to access. It may be necessary to advocate strongly to state government departments to gain both this assessment and the committed involvement of behavioural support teams; to do otherwise puts the patient at risk of ineffective, expedient solutions. Some important syndrome specific morbidities are summarised in *Table 2*.

Resources

A recent website specifically for primary care physicians provides excellent information at: www.ddhealthinfo.org/ggrc

A more comprehensive website with a focus on genetic conditions is Online Mendelian Inheritance in Man at: www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM

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