Australian Government

Department of Health and Aged Care

Annual Health Assessment for People with Intellectual Disability

YOUNG PERSON (12 to 18 years)

Name of the Young Person (having the assessment)

This document is reviewed annually and updated to be consistent with current relevant evidence and national and international guidelines. Check the Department of Health and Aged Care website for the latest version of this document.

For more information and resources on annual health assessments for people with intellectual disability visit www.health.gov.au/ahas or email Intellectualdisability@health.gov.au.

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People supporting the annual health assessment

Name of Family Member / Carer / Support Worker

Name of General Practitioner (GP)

Steps to follow

1. You (the person with intellectual disability), a family member and/or support staff fill in **Part 1** of this book. It is OK to get information from records, family members and staff.
2. Make a long appointment with your usual GP.
   - Ask the receptionist if the clinic has a Practice Nurse, as they may be able to assist with the health assessment.
   - You may wish to book the first appointment of the day to avoid long waiting times.
3. Take this book to the appointment with the GP and/or email it to your GP before the appointment.
4. The GP will look through the first part of this book and fill in **Part 2** while examining you.
5. It is important that the Action Plan is filled in at the end of the consultation. A copy of the Action Plan is to be kept with your personal record.
6. Medicare supports this ANNUAL health assessment. Please refer to Page 19 for details of Medicare items.

People with intellectual disability generally have poor health. Their healthcare is generally of a lower standard. The Comprehensive Health Assessment Program (CHAP) was designed to improve the interaction between the GP and the person with intellectual disability.

It does this by:

- recording a dedicated health history from you and/or your carer
- improving communication between, and integration of, disability and health services
- educating and empowering all those involved in the health assessment
- providing guidelines for the GP on commonly missed health issues for this population.

Use of the CHAP can improve your health. Research (including several randomised controlled trials) has demonstrated its benefit in detecting new disease, and enhancing health screening and promotion.

Part 1

To be completed by you, your family and/or support staff.

Tick the boxes below if you have experienced any of the following signs and symptoms in the **last year**.

If you are unsure or don't know the answer, please tick the 'unsure/don't know' box.

To make an accurate medical assessment of your health, the doctor needs to know about these signs and symptoms.

1. Breathing

	Yes	No	Unsure/ don't know
Do you **cough**?	☐	☐	☐
Do you cough up **blood**?	☐	☐	☐
Do you have a **runny** nose?	☐	☐	☐
Do you cough up **stuff/mucous/sputum**?	☐	☐	☐
Do you get short of **breath**?	☐	☐	☐
Do you **wheeze**?	☐	☐	☐

2. Heart

	Yes	No	Unsure/ don't know
Do you have a **heart disease** that you have had from birth?	☐	☐	☐
Do you have **chest pain**?	☐	☐	☐
Does your heart 'race'/**beat quickly**?	☐	☐	☐
Do your **ankles swell**?	☐	☐	☐
Do you get **short of breath** while lying in bed?	☐	☐	☐
Do you get a **blue tinge to your skin** (for example, fingers/lips/toes)?	☐	☐	☐

3. Muscles and Joints

	Yes	No	Unsure/ don't know
Do you have **joint pain or back pain**?	☐	☐	☐
Do you have **muscle pain or weakness**?	☐	☐	☐
Do you use **walking aids or orthotics**?	☐	☐	☐
Do you have **scoliosis** (curved or twisted backbone) **or contractures** (painful tight muscles)?	☐	☐	☐

4. Stomach/Bowel

	Yes	No	Unsure/ don't know
Have you **lost weight**?	☐	☐	☐
Do you have **trouble swallowing**?	☐	☐	☐
Do you **regurgitate/vomit**?	☐	☐	☐
Do you get **heartburn**?	☐	☐	☐
Do you have **diarrhoea**?	☐	☐	☐
Do you have **black bowel motions/faeces/poo**?	☐	☐	☐
Do you get **constipated**?	☐	☐	☐
Do you lose control of **bowel motions**?	☐	☐	☐
Do you have **abdominal/stomach pain**?	☐	☐	☐

	Yes	No	Unsure/ don't know

5. Urinary system

	Yes	No	Unsure/ don't know
Do you have **pain** when passing **urine/wee**?	☐	☐	☐
Do you have **blood** in your **urine**?	☐	☐	☐
Do you **lose control** of your **urine**?	☐	☐	☐
Can you take care of your **toileting needs**?	☐	☐	☐
Do you **urinate more** than in the past?	☐	☐	☐

6. Nervous system

Questions about epileptic episodes (fits) are covered in section 13.

	Yes	No	Unsure/ don't know
Do you **have headaches**?	☐	☐	☐
Do you **faint**?	☐	☐	☐
Do you get **unsteady** when walking?	☐	☐	☐
Do your arms or legs become **weak**?	☐	☐	☐
Do you have **tingling** or strange feelings in your skin?	☐	☐	☐

7. Skin

	Yes	No	Unsure/ don't know
Do you have **any skin disease or rash**?	☐	☐	☐

8. Pain

How do you react to pain?

How do you communicate pain?

9. Medications

It is important for the doctor to know about ALL your medication.
List the medication/s prescribed by any doctor.

Name	How often and how much is taken?	When was it first prescribed?
		DD/MM/YYYY
		DD/MM/YYYY
		DD/MM/YYYY
		DD/MM/YYYY
		DD/MM/YYYY
		DD/MM/YYYY
		DD/MM/YYYY
		DD/MM/YYYY

When were medications last reviewed?

Who reviewed the medications?

List any medications bought 'over the counter' without a script, including alternative health remedies.

Name	How often and how much is taken?

10. Allergies

Please list anything you are allergic to, including medications.

What are you allergic to?

	Yes	No	Unsure/ don't know

11. Sleep

Do you experience any problems with **sleeping**?	☐	☐	☐
If **Yes**, please describe			

12. Cause of intellectual disability

Is the cause of your intellectual disability known?	☐	☐	☐
If **Yes**, please give details.			

13. Epilepsy

Have you ever had epileptic seizures/fits?	☐	☐	☐

If **No** or **Unsure**, please go to **question 14**

If **Yes**, please describe and list the **type, number** and **usual duration** of seizures during the last year.

Type of seizures	Number	Usual duration

Which doctor treats the epileptic seizures/fits?	Please tick one
GP	☐
Neurologist	☐
Other Please describe.	☐

Epilepsy – *Continued*

When did this doctor last review your epilepsy?		
Month and year if known MM/YYYY Unknown ☐		
Since this review, have the seizures	Please tick as applicable	
Become more severe?	☐	
Become more frequent?	☐	
Remained the same?	☐	
Improved?	☐	
Do you have an Epilepsy Management Plan?	Yes ☐	No ☐

14. Sexual orientation and gender identity

What is your gender identity?	Please tick one
Male	☐
Female	☐
Non-binary	☐
Other	☐
I do not want to answer	☐

What sex was originally listed on your birth certificate?	Please tick one
Male	☐
Female	☐
Other Please describe.	☐

Do you identify as	Please tick as applicable
Straight or heterosexual	☐
Bisexual	☐
Lesbian or gay	☐
Other	☐
I do not want to answer	☐

	Yes	No	Unsure/ don't know

15. Relationships

The information you provide must be kept confidential.

You do not have to answer these questions if they make you uncomfortable, but it is very important you tell your GP about any concerns you have.

	Yes	No	Unsure/ don't know
Have you ever been sexually active?	☐	☐	☐
Have you ever been abused (sexual, physical or psychological)?	☐	☐	☐
Comments			

For young men and people with a penis, please go to question 17

16. Health of women and people with a cervix

	Yes	No	Unsure/ don't know
Are you concerned that you reached puberty too early or too late?	☐	☐	☐
Do you have menstrual periods?	☐	☐	☐
Do you care for your periods yourself?	☐	☐	☐
Do you use any form of contraception?	☐	☐	☐
If **Yes**, please describe.			
Date of last period (if any) DD/MM/YYYY			

Do you have any of the following:	Yes	No	Unsure/ don't know
Premenstrual Syndrome?	☐	☐	☐
Irregular periods?	☐	☐	☐
Heavy periods?	☐	☐	☐
Painful periods?	☐	☐	☐
Abnormal vaginal discharge?	☐	☐	☐
Vaginal irritation (for example, thrush)?	☐	☐	☐
Pelvic pain?	☐	☐	☐

For women and people with a cervix, please go to question 18

17. Health of young men and people with a penis

	Yes	No	Unsure/ don't know
Do you have any problems with your penis (for example, sores, scars or discharge)?	☐	☐	☐
Are both of your testes in the scrotum?	☐	☐	☐
Are you concerned that you reached puberty too early or too late?	☐	☐	☐
Do you have any erection dysfunction?	☐	☐	☐

The remaining questions are for all young people

	Yes	No	Unsure/ don't know

18. Behaviours of concern

Do you have any behaviours of concern?	☐	☐	☐
If **Yes**, please describe the behaviours, any consequence (legal or otherwise), and any help you receive for these behaviours.			

19. Mental health

Do you have any mental health problems?	☐	☐	☐
If **Yes**, please describe the mental health problem/s and any help you receive for the problem/s.			

	Yes	No	Unsure/ don't know

20. Seeing

Do you have a known problem with seeing?	☐	☐	☐
If **Yes**, please describe the problem.			
If **No** or **Unsure/don't know**			
Are you or other people concerned you have a problem with seeing?	☐	☐	☐
Have you ever been prescribed glasses?	☐	☐	☐
If **Yes**, do you usually wear the glasses?	☐	☐	☐

When was the last vision test?

Month and year if known MM / YYYY

Unknown ☐

Never ☐

Who performed this test?	Please tick one
Eye doctor / ophthalmologist	☐
Optometrist	☐
GP	☐
Test while at school	☐
Unknown	☐

Result of last vision test	Please tick one
Normal	☐
Unknown	☐
Abnormality found	☐

If an abnormality was found, please describe.

	Yes	No	Unsure/ don't know

21. Hearing

Do you have a known problem with hearing?	☐	☐	☐
If **Yes**, please describe the problem.			
If **No** or **Unsure/don't know**			
Are you or other people concerned you have a problem with hearing?	☐	☐	☐
Have you been prescribed a hearing aid?	☐	☐	☐
If **Yes**, do you usually wear the hearing aid?	☐	☐	☐
Do you use any other hearing device?	☐	☐	☐

When was your last hearing test?

Month and year if known MM / YYYY

Unknown ☐

Never ☐

Who performed this test?	**Please tick one**
Audiologist	☐
GP	☐
Other	☐

Result of last hearing test	**Please tick one**
Normal	☐
Unknown	☐
Other	☐

If an abnormality was found, please describe.

22. Health promotion and screening

Dental

Date of last review DD/MM/YYYY

Unknown ☐

Blood pressure

Date of last check DD/MM/YYYY

Unknown ☐

Cigarettes smoked (per day)

Alcohol (standard drinks per week)

Recreational drugs (list any used)

Thyroid function test (TFT)

If you have Down syndrome, when was the most recent test? DD/MM/YYYY

Unknown ☐

Vitamin D test

Vitamin D deficiency appears to be quite common in people with intellectual disability.

Date of last review DD/MM/YYYY

Unknown ☐

Diabetes test

Date of test DD/MM/YYYY

Unknown ☐

Skin check

Date of last review DD/MM/YYYY

Unknown ☐

	Yes	No	Unsure/ don't know

23. Activity and lifestyle

	Yes	No	Unsure/ don't know
Have you noticed any mobility changes over time?	☐	☐	☐
Has there been any decline in your abilities, (for example, memory, dealing with money, making meals)?	☐	☐	☐
Are you physically active?	☐	☐	☐
Do you have any abnormal eating behaviours?	☐	☐	☐
Do you or others have concerns about the quality of your diet?	☐	☐	☐
Inactivity and poor diet can lead to significant health problems and depression.			

24. Immunisations

Please fill in the following about your immunisations.

Ask your GP if there are any immunisations you should get, either as a booster or for the first time.

	Yes	No	Unsure/ don't know
COVID-19 Is your COVID-19 immunisation up to date? If **Yes**, date/s of last immunisation/s DD/MM/YYYY DD/MM/YYYY DD/MM/YYYY If **No**, reason why	☐	☐	☐
Tetanus/Diphtheria/Pertussis (Whooping Cough) If **Yes**, date of most recent immunisation DD/MM/YYYY If **No**, reason why	☐	☐	☐
Hepatitis A If **Yes**, date of most recent immunisation DD/MM/YYYY If **No**, reason why	☐	☐	☐

	Yes	No	Unsure/ don't know

Immunisations – *continued*

	Yes	No	Unsure/ don't know
Hepatitis B If **Yes**, date of most recent immunisation DD / MM / YYYY If **No**, reason why	☐	☐	☐
Influenza If **Yes**, date of most recent immunisation DD / MM / YYYY If **No**, reason why	☐	☐	☐
Pneumococcus If **Yes**, date of most recent immunisation DD / MM / YYYY If **No**, reason why	☐	☐	☐
Measles, Mumps and Rubella If **Yes**, date of most recent immunisation DD / MM / YYYY If **No**, reason why	☐	☐	☐
Meningococcal meningitis If **Yes**, date of most recent immunisation DD / MM / YYYY If **No**, reason why	☐	☐	☐
HPV (Human Papilloma Virus) If **Yes**, date of most recent immunisation DD / MM / YYYY If **No**, reason why	☐	☐	☐

	Yes	No	Unsure/ don't know

25. Developmental concerns

	Yes	No	Unsure/ don't know
Do you, your family or carers have any concerns about your development, such as problems with speech and communication, fine motor skills (for example, using hands), gross motor skills (for example, walking), self-care, social skills and learning or school problems?	☐	☐	☐
If **Yes**, please describe the problem/s.			
Do you see any allied health professionals (for example, speech therapist)?	☐	☐	☐
If **Yes**, please give details.			

26. Social situation

	Yes	No	Unsure/ don't know
Who do you live with?			
Name of your school and grade/level			
Who has parental responsibility for you (for example, parent, carer, child protection)?			
Do you live in residential care or out of home care (for example, foster care, youth housing service, supported independent living)?	☐	☐	☐
Do you receive NDIS support?	☐	☐	☐
Are there any concerns about this support?	☐	☐	☐
Do your family receive a carer allowance?	☐	☐	☐

27. Family medical history

Has anyone in your family (blood relation) had any of the following conditions?	Yes	No	Unsure/ don't know
Diabetes	☐	☐	☐
Mental health problems	☐	☐	☐
Heart disease	☐	☐	☐
Epilepsy	☐	☐	☐
Breast cancer	☐	☐	☐
Intellectual disability	☐	☐	☐
Bowel cancer	☐	☐	☐

28. Your medical history

List any information about your health history that has not been recorded in this booklet. Include any medical problems, surgery/operations, gynaecological, and mental health problems.

29. Summary of health concerns

List the concerns you, your family and/or carers have about your health.

30. Person who completed Part 1 of this document

Name

Date completed

DD / MM / YYYY

Remember!

If the clinic has a Practice Nurse, speak with them about completing this assessment as they may be able to assist. This will make the consultation with the GP easier.

Part 2

To be completed by the General Practitioner.

Thank you for reviewing this young person's health care.

Date of visit

DD / MM / YYYY

Please review the history provided in Part 1 of this document.

Common health deficits for people with intellectual disability include:

- unrecognised pain or infections
- poor dental care
- over-use and inadequate review of anti-psychotics/anti-convulsants
- unrecognised mental disorder/s, poor psychiatric assessment / management
- inadequate epilepsy assessment / management
- unrecognised / poorly managed constipation
- unrecognised / poorly managed dysphagia and aspiration
- unrecognised reflux esophagitis / H.pylori infection
- unrecognised or poorly managed hearing and vision impairment +/- pathology
- incomplete immunisation schedules
- incomplete health screens – BP, skin, breast, cervical
- unrecognised osteoporosis and Vitamin D deficiency
- unrecognised or poorly managed obesity
- precocious/delayed puberty, delayed menstruation, growth issues
- undescended testes/ hypogonadism
- inadequate information about menstrual management
- inadequate information about human relations and sexuality
- uninvestigated cause of intellectual disability.

The Medicare Benefits Schedule items for Health Assessments provided for people with intellectual disability are:

- 701 (brief)
- 703 (standard)
- 705 (long) or
- 707 (prolonged)

A health assessment for people with intellectual disability may be claimed once every twelve months by an eligible patient.

The Chronic Disease Care Management items are:

- GP Management Plan (item 721)
- Team Care Arrangements (item 723)

Please perform a COMPREHENSIVE REVIEW of your patient's health:

- full physical examination
- vision and hearing screening
- urine analysis
- other tests you feel are indicated.

Please record your findings on the following pages.

There is syndrome specific information for you to keep at the end of this booklet.

Using this Comprehensive Health Assessment Program (CHAP) can improve the health of a person with intellectual disability. A randomised controlled trial has demonstrated its beneficial effect to detect new disease, and enhance health screening and promotion. This information is updated on an annual basis in line with current evidence and national and international guidelines.

1. Check

Height

Check against **growth chart**.

Growth charts are available on the RCH and ANZSPED websites. ANZSPED also has specific growth charts for Down syndrome.

Height		cm
Weight		kg
Waist measurement (if necessary)		cm
Blood pressure		mmHg
Pulse rate		BPM

If the person has a weight / blood pressure problem, please specify action recommended or taken:

Weight control	
Hypertension	

2. Systems check

Please perform a complete comprehensive physical examination.

	New findings
Cardiovascular	
Respiratory	
Musculo-skeletal	
Renal / urogenital	
Endocrine	
Gastrointestinal	
Nervous	
Skin	

3. Mental health problems

Common problems are adjustment to transitions / grief, risk of neglect / abuse, anxiety disorders, mood disorders (for example, depression), emerging psychosis.

Is there any evidence of a mental health problem? Yes ☐ No ☐

If **Yes**, please describe.

Is there any evidence of self harm, suicidal thought or thoughts of harming others? Yes ☐ No ☐

If **Yes**, please describe.

4. Epilepsy

If present

Has the person's seizure control been reviewed? Yes ☐ No ☐

Does the person have an epilepsy management plan? Yes ☐ No ☐

Has the risk of SUDEP been minimised? Yes ☐ No ☐

Has a referral been considered? Yes ☐ No ☐

5. Vision test

For those who cannot name letters, please use the last page to aid testing

Has the person's vision been tested? Yes ☐ No ☐

Test result

Both eyes ______ Right ______ Left ______

Unable to test ☐ Uncertain ☐

If abnormal, unable to test or uncertain, consider referral.

6. Hearing test

Appropriate hearing test: Whisper test both sides at 0.6 metre and tympanometry

Has a hearing test been performed? Yes ☐ No ☐

Test result

Right ear ______ Left ear ______

Unable to test ☐ Uncertain ☐

If abnormal, unable to test or uncertain, consider referral.

Has an otoscopy been performed? Yes ☐ No ☐

If hearing test or otoscopy abnormal, unable to test or uncertain, consider referral.

7. Aetiology

If there is no definitive diagnosis and the tests below have not been performed

Discuss genetic testing covering the potentials advantages and disadvantages of pursuing an underlying genetic diagnosis

a advantages: support groups, assist NDIS funding, reproductive confidence, an explanation, link to specific management guidelines

b disadvantages (occasionally): currently limited treatments, adjustment to a diagnosis, chance of test being non diagnostic and giving uncertain or incidental findings.

Recommend a chromosome microarray AND Fragile X PCR (Medicare indication: Intellectual disability).

If regression of skills or multi-organ involvement, order a urinary metabolic screen.

If there is an underlying genetic cause identified, then discuss if the person would like a referral to a genetic counsellor to help them understand their genetic condition and what this means for their care and family planning.

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8. Sleep

Does the person have sleep apnoea or sleep problems? Yes ☐ No ☐

9. Sexual development and urogenital health

Recommend a review of their reproductive health, sexual activity and sexual development.

Has the person developed age appropriate sexual characteristics? Check Tanner score (for example, Australian Family Physician (2017) 46: 913) Yes ☐ No ☐

If **No**, what action was taken?

Was the young person checked for undescended testicles? Yes ☐ No ☐

If **Yes**, what abnormalities were found?
Action taken

10. Abuse and Trauma

Check for signs of physical, psychological or sexual abuse and trauma.

11. Dental Health

Is there obvious dental pathology? Yes ☐ No ☐

Has the person been reviewed by a dentist in the last six months? Yes ☐ No ☐

Does the person need dental assessment? Yes ☐ No ☐

12. GIT and bladder function

Especially for people with cerebral palsy

Has the person been assessed for dysphagia? Yes ☐ No ☐

Has the person been assessed for gastro-oesophageal disease? Yes ☐ No ☐

Does the person experience urinary or bowel incontinence? Yes ☐ No ☐

Does the person have chronic constipation? Yes ☐ No ☐

13. Activity and lifestyle

Inactivity and poor diet can lead to significant health problems and depression

Does the person have sufficient **exercise**? Yes ☐ No ☐

Does the person have any **diet problems** / **abnormal eating behaviours**? Yes ☐ No ☐

Is a referral needed for any of these? Yes ☐ No ☐

14. Immunisation status

Are the following immunisations up to date?

	Yes	No
COVID-19	Yes ☐	No ☐
Tetanus, diphtheria and pertussis (dTpa)	Yes ☐	No ☐
Booster indicated	Yes ☐	No ☐
Booster given	Yes ☐	No ☐
Hepatitis A	Yes ☐	No ☐
Hepatitis B	Yes ☐	No ☐
Influenza	Yes ☐	No ☐
Pneumococcus	Yes ☐	No ☐
Measles, Mumps and Rubella	Yes ☐	No ☐
Meningococcal	Yes ☐	No ☐
HPV cervical cancer	Yes ☐	No ☐

Influenza, Pneumococcus and Meningococcal immunisation may be indicated for people with intellectual and developmental disability. For more details, please refer to The Australian Immunisation Handbook: https://immunisationhandbook.health.gov.au/

15. Health promotion and screening

	Yes	No
Blood glucose – needs to be tested?	Yes ☐	No ☐
Lipid Screen – needs to be tested?	Yes ☐	No ☐
Thyroid function test (Down syndrome) – needs to be tested?	Yes ☐	No ☐
Vitamin D (if on anti-epileptic medication) – needs to be tested?	Yes ☐	No ☐

16. Medication review

Including prescription and non-prescription medications

	Yes	No
Have the person's medications been reviewed?	Yes ☐	No ☐
Have you communicated indications, side effects and interactions?	Yes ☐	No ☐

17. Future planning

	Yes	No
Have there been any **mobility** changes over time?	Yes ☐	No ☐
Has there been any **functional decline** over time?	Yes ☐	No ☐
Would you be surprised if the person were to die in the next 12 months?	Yes ☐	No ☐
If **No**, have you considered **advanced care planning and palliative care**, and discussed with the person, their family or carers?	Yes ☐	No ☐

Please assist the young person, carer or support worker to complete the 'ACTION PLAN' on the following page. A copy of the ACTION PLAN can be given to the person and their care provider. This book can be returned to the care provider who came to this consultation.

Thank you for your comprehensive health review of this person. Your efforts are part of improving the poor health status of the 450,000+ Australians with intellectual disability.

Based on this, you can make a further appointment to do a formal management plan.

The Medicare Benefits Schedule items for Health Assessments provided for people with intellectual disability are:

- 701 (brief)
- 703 (standard)
- 705 (long) or
- 707 (prolonged).

The Chronic Disease Care Management items are:

- GP Management Plan (item 721)
- Team Care Arrangements (item 723)

Thank you for your assistance.

Name of person

Address

ACTION PLAN
What actions to do next and by whom

Problem(s) identified	**Action(s) to be taken, including medication changes** (for medication changes, name medication, how often, how much OR whether to cease)	**Action(s) by, or arranged by whom**	**By when (date)**	**Next review (date)**
			Done ☐	
			Done ☐	
			Done ☐	
			Done ☐	

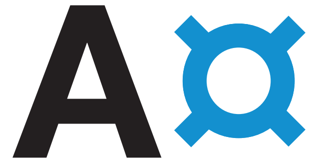
Completed by (please print)

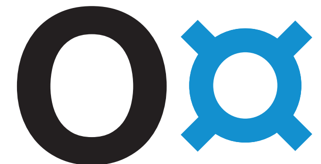
Signature

Syndrome specific list for general practitioners					
SYSTEM	**CEREBRAL PALSY 1:500**	**DOWN SYNDROME 1:700**	**PRADER-WILLI 1:25,000**	**FRAGILE X 1:6000**	**PHENYL/KETONURIA 1:10 000-1:20000**
AUDIOVISUAL	Visual Impairment Hearing Impairment	Visual impairment (multifactorial), cataracts Hearing impairment (multifactorial) (annual assessments recommended)	Strabismus Myopia	Visual Impairment (multifactorial) Hearing Impairment Recurrent ear infections	
ENDOCRINE		Hypothyroidism (annual TFT recommended)	NIDDM (secondary to obesity) Hypogonadism Delayed puberty		
PSYCHIATRIC/ PSYCHOLOGICAL	Depression Variable intellectual capacity	Depression Alzheimer's type dementia (clinical onset uncommon before 40 years)	Hyperphagia Impulse control difficulties Self-injury	Attention deficit/ hyperactivity Variable intellectual capacity Disabled in social functioning	Variable intellectual capacity Phobic anxiety Disabled in social functioning
C.N.S.	Epilepsy	Epilepsy Usually clonic/tonic		Epilepsy Usually clonic/tonic, complex partial	Epilepsy Hyperactivity Tremor and pyramidal tract signs Extrapyramidal syndromes
CARDIOVASCULAR		Congenital Heart Defects (common – in 40 to 50%)		Aortic dilatation, Mitral Valve prolapse (related to connective tissue dysplasia)	
MUSCULR/ SKELETAL AND SKIN	Orthopaedic problems Neuromuscular problems	Atlantoaxial instability Skin disorders, alopecia, eczema	Scoliosis, Kyphosis Hypotonia Skin picking	Connective tissue dysplasia Scoliosis Congenital Hip Dislocation	
OTHER	Genito-urinary problems Incontinence Constipation Dental problems Recurrent aspiration Oesophagitis, gastroesophageal reflux +/- bleeding/anaemia Swallowing/eating difficulties	Blood dyscrasias childhood leukaemia Sleep apnoea Increased susceptibility to infections, Coeliac disease	Infantile failure to thrive, then hyperphagia and severe obesity High tolerance to pain Decreased ability to vomit Sleep apnoea Osteoporosis Undescended testes Dental Abnormalities	Herniae (CT related) Abnormalities of speech and language	Eczema
INHERITANCE		Most cases are sporadic; 4% due to translocation involving chromosome 21 or rarely parental mosaicism	Atypical. Most cases are sporadic.	X linked	Autosomal recessive

Syndrome specific list for general practitioners						
SYSTEM	**ANGELMANN SYNDROME <1:10 000**	**WILLIAMS ≤1:20 000**	**RETT 1:14 000 FEMALES**	**NOONAN <1:10 000**	**TUBEROUS SCLEROSIS 1:6 000-17 000**	**NEUROFIBRO MATOSIS 1:3000**
AUDIOVISUAL	Glaucoma	Hyperacusis Strabismus	Refractory errors	Strabismus, refractive errors Vision/hearing impairments	Retinal tumours Eye rhabdomyomata	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
C.N.S.	Severe developmental delay Epilepsy	Perceptual & 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	Rhabdomyomata Hypertension	
MUSCULAR / SKELETAL	Joint contractures and scoliosis (in adults)	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 the 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	Usually sporadic X linked	Autosomal dominant may be sporadic	Autosomal dominant	Autosomal dominant

Adapted from an original unpublished version by Michael Kerr and Glyn Jones.





If the young person has difficulty naming letters from the Snellen Chart, ask them to point to the corresponding letter on this page.

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All information in this publication is correct as at April 2024