Australian Government Department of Health and Aged Care





# Annual Health Assessment for People with Intellectual Disability ADULT

Name of Person (with intellectual disability)

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

For more information on how the Australian Government is improving annual health assessments for people with intellectual disability visit <a href="https://www.health.gov.au/our-work/improving-annualhealth-assessments-for-people-with-intellectual-disability">https://www.health.gov.au/our-work/improving-annualhealth-assessments-for-people-with-intellectual-disability</a>.

## People supporting the annual health assessment

Name of Family Member / Support Staff

Name of General Practitioner (GP)

## **Steps to follow**

- 1. The person with disability, family member and/or support staff fills in **Part 1** of this book for the person with disability. It is OK to get information from records, family members and staff.
- 2. Make a long appointment for the person with disability with their 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.
- 4. The GP will look through the first part of this book and fill in Part 2 while examining the individual.
- 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 the individual's personal record.
- 6. Medicare supports this ANNUAL health assessment. Please refer to Page 17 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 the person with disability and or their 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 the health of a person with intellectual disability.

Several randomised controlled trials and observational studies have demonstrated its benefit in detecting new disease, enhanced health screening and promotion.

## Part 1 – For the Person with Disability

Please tick the boxes below if the person has 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 the person's health, the doctor needs to know about these signs and symptoms.

		Yes	Νο	Unsure/ don't know
1.	Breathing system			
	Does the person <b>cough</b> ?			
	Does the person cough up <b>blood</b> ?			
	Does the person cough up <b>stuff/mucous/sputum</b> ?			
	Does the person get short of <b>breath</b> ?			
	Does the person <b>wheeze</b> ?			
2.	Heart system			
	Does the person have <b>chest pain</b> ?			
	Does the person's heart 'race'/ <b>beat quickly</b> ?			
	Do the person's <b>ankles swell</b> ?			
	Does the person get <b>short of breath</b> while lying in bed?			
	Does the person get a <b>blue tinge to their skin</b> (for example, fingers/lips/toes)?			
3.	Muscles & Joints			
	Does the person have joint pain or <b>back pain</b> ?			
	Does the person have <b>muscle pain</b> ?			
4.	Gastrointestinal system			
	Has the person <b>lost weight</b> ?			
	Does the person have <b>trouble swallowing</b> ?			
	Does the person regurgitate/vomit?			
	Does the person get <b>heartburn</b> ?			
	Does the person have <b>diarrhoea</b> ?			
	Does the person have <b>black bowel motions/faeces</b> ?			
	Does the person get <b>constipated</b> ?			
	Does the person lose control of <b>bowel movements</b> ?			
	Does the person have <b>abdominal/stomach pain</b> ?			
5.	Urinary system			
	Does the person have <b>pain</b> when <b>urinating</b> ?			
	Does the person have <b>blood</b> in the <b>urine</b> ?			
	Does the person <b>urinate involuntarily</b> /suffer incontinence?			

Does the person urinate **a lot**/more than usual?

		Unsure/
		don't
Yes	No	know

#### 6. Nervous system

Please note that epileptic episodes are not included here.

Does the person <b>faint</b> ?		
Does the person get <b>unsteady</b> when walking?		
Do the person's arms or legs become <b>weak</b> ?		
Does the person have <b>tingling</b> or strange feelings in the skin?		

#### 7. Pain perception

How does the person react to pain?

How does the person communicate pain?

#### 8. Medications

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

Name	How often and how much is taken?

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

Name	How often and how much is taken?

### 9. Allergies

Please list anything the person is allergic to, including medications.

		Unsure/ don't
Yes	No	know

## 10. Sleep

Does the person experience any problems with sleeping?		
If <b>Yes</b> , please describe	·	

## 11. Cause of intellectual disability

Is the cause of intellectual disability known?		
If <b>Yes</b> , please describe		

## 12. Epilepsy

Has the person ever had epileptic seizures/fits?		
If <b>No</b> , please go to Question 13		
If <b>Yes</b> , please describe		

Type of Seizures	Number	Usual duration

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

When did this doctor last review the person's 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	

## 13. Sexual orientation and gender identity

What is the person's gender identity:	Please tick one
Male	
Female	
Non-binary	
Decline to answer	
Other	

What sex was originally listed on the person's birth certificate?	Please tick one
Male	
Female	
Other Please describe	

Does the person identify as	Please tick one
Straight or heterosexual	
Bisexual	
Lesbian or gay	
Decline to answer	
Other	

#### 14. Relationships

Has the person ever been sexually active?		
Has the person ever been abused (sexual, physical or psychological)?		
Comments		

#### For men and people with a penis, please go to question 16

#### 15. Health of women and people with a cervix

Does the person have menstrual periods?		
If <b>Yes</b> , date of last period DD/MM/YYYY Unknown		
Does the person use Implanon?		
Does the person use an IUD (intrauterine device)? Sometimes these are called 'Mirena' or 'Kyleena'		
Does the person take the oral contraceptive pill?		
If <b>Yes</b> , what is the name of the pill:		
Does the person use any other contraceptive method?		
If <b>Yes</b> , please describe:	·	

Does the person have any of the following:		
Pre-Menstrual Syndrome?		
Mid-cycle bleeds?		
Painful periods?		
Abnormal vaginal discharge?		
Vaginal irritation (e.g. thrush)?		
Pelvic pain?		
Menopausal symptoms (e.g. hot flushes)?		

Women and people with a cervix with intellectual disability have the same reason for needing cervical screening as those in the general population. It is recommended for both human papillomavirus (HPV) vaccinated and unvaccinated people, that an HPV test should be performed every five years starting at 25 years of age regardless of sexual orientation.

Since 1 July 2022, all people can be offered a choice of HPV testing either through a self-collected vaginal sample at the person's general practice or a clinician-collected sample from the cervix.

Taking a self-collected vaginal sample may be assisted by a GP, if necessary, or if the GP is confidant a good sample can be collected, it can be taken by the person in a private place without assistance. This change is likely to make cervical screening accessible to more people with a disability.

#### Health of women and people with a cervix – Continued

Has the person had an HPV test?		
If <b>Yes</b> , date of last test DD/MM/YYYY Unknown		
Has the person had a cervical screening?		
If <b>Yes</b> , date of last screening DD/MM/YYYY Unknown		
Does the person need cervical screening?		

If screening is needed but has not happened, please indicate why:

Distress about the procedure	
Screening planned	
Desensitisation/preparation planned	
Desensitisation/preparation in progress	
Other	

A mammogram should be arranged every 2 years for women and people with a cervix who are over 50 years of age, or more regularly for people with a family history of breast/ovarian cancer, or Ashkenazi Jewish ancestry.

Are the person's breasts checked by a GP?		
Has the person ever had a mammogram?		
If <b>Yes</b> , date of last mammogram DD/MM/YYYY Unknown		

If a mammogram is needed but has not happened, please indicate why:

Distress about the procedure	
Mammogram planned	
Desensitisation/preparation planned	
Desensitisation/preparation in progress	
Other	

#### For women and people with a cervix, please go to question 17

#### 16. Health of men and people with a penis

Does the person have any genital abnormality or discharge from their penis?		
Does the person have any erection dysfunction?		
Does the person have undescended testicles?		

#### Please go to question 17

		Unsure/
		don't
Yes	No	know

#### 17. Behaviours of concern

Does the person have any behaviours of concern?			
If <b>Yes</b> , please describe the behaviours and any support the person receive behaviours.	es to mana	age these	
8. Mental health			
Does the person have any mental health problems?			
If Yes, please describe the problem and any help the person receives to r	nanage thi	S.	

	Yes	Νο	Unsure don't know
<b>/</b> ision			
Does the person have a known problem with vision?			
If <b>Yes</b> , please describe the problem.			
If <b>No</b> or <b>Unsure/don't know</b>			
Do you suspect the person may have a problem with vision?			
Has the person ever been prescribed glasses?			
If <b>Yes</b> , does the person usually wear the glasses?			
When was the last vision test?	L.	l	
Unknown Never			
Who performed this test?	Please tic	k one	
Eye doctor / ophthalmologist			
Optometrist			
GP			
Test while at school			
Unknown			
Result of last vision test	Please tic	k one	
Normal			
Unknown			
Abnormality found			
If an abnormality was found, please describe			

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		Yes	No	Uns do kn
ave a known problem w	ith hearing?			
escribe the problem.				
don't know				
the person may have a	problem with hearing?			
been prescribed a hear	ing aid?			
erson usually wear the h	nearing aid?			
year if known MM/ ] ]				
his test?		Please tio	ck one	
ring test		Please tio	sk one	
/ was found, please des	scribe.			

#### 21. Health promotion & screening

#### Dental

Date of last review

Unknown 🗌

Unknown

D D	/ M	M / Y	YYY	

#### **Blood pressure**

Date of last check

D D / M M / Y Y Y Y

#### Cigarettes smoked (per day)

Alcohol (standard drinks per week)

### Thyroid function test (TFT)

If the person has Down syndrome, what was the date of last test? Unknown

D D / M M / Y Y Y Y

#### Vitamin D test

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

Date of last review	D D / M M / Y Y Y Y

Unknown 🗌

#### **Bowel cancer test**

Current guidelines recommend that people aged over 50 years should complete a bowel cancer screening test known as a Faecal Occult Blood Test every two years. People may also need a bowel cancer assessment if they have a family history of bowel cancer, a personal history of inflammatory bowel disease, or symptoms of bowel cancer or with high-risk familial syndromes, such as Lynch syndrome or familial polyposis.

Date of test

D D / M M / Y Y Y Y	/ Y Y Y Y
---------------------	-----------

Unknown

Date of test

Unknown	
---------	--

D D / M M / Y Y Y Y
---------------------

#### Skin check

Date of last review DD/MM/Y

Unknown 🗌

	/	$/ \vee$	

## 22. Activity & lifestyle

Has there been any mobility changes over time?					
Has there been any decline in the person's abilities, (for example, memory, dealing with money, making meals)?					
Is the person physically active?					
Does the person have any abnormal eating behaviours?					
Are there concerns about the quality of the person's diet?					
Inactivity and poor diet can lead to significant health problems and depression.					

#### 23. Immunisations

<b>COVID-19</b> Is the person's COVID-19 immunisation up to date?				
If <b>Yes</b> , date/s of last imm	 1	1		
DD/MM/YYYY	<b>D</b> D / M M / Y Y Y Y	D D / M M / Y Y Y Y		
lf <b>No</b> , reason why				
Tetanus/Diphtheria/Pertu	ssis			
If Yes, date of most rece	nt immunisation			
DD/MM/YYYY				
If <b>No</b> , reason why				
Hepatitis A If required - check with the GF				
If Yes, date of most rece	nt immunisation		 ·	
DD/MM/YYYY				
If <b>No</b> , reason why				
Hepatitis B				
If required - check with the GF If <b>Yes</b> , date of most rece				
DD/MM/YYYY				
If <b>No</b> , reason why				

	Yes	No	Unsure/ don't know
Immunisations – continued			
Influenza (If required – check with the GP)			
If Yes, date of most recent immunisation			
DD/MM/YYYY			
If <b>No</b> , reason why			
Pneumococcus (If required – check with the GP)			
If Yes, date of most recent immunisation			
D D / M M / Y Y Y Y			
If <b>No</b> , reason why			
Measles, Mumps and Rubella (If required – check with the GP)			
If Yes, date of most recent immunisation		1	
D D / M M / Y Y Y Y			
If <b>No</b> , reason why			
Meningococcal Meningitis (If required – check with the GP)			
If Yes, date of most recent immunisation			
D D/M M/Y Y Y Y			
If <b>No</b> , reason why			
Cervical Cancer			
(If required – check with the GP) If <b>Yes</b> , date of most recent immunisation			
If <b>No</b> , reason why			
Shingles/Herpes Zoster (If 60 years or over – check with the GP as cost to client)			
If <b>Yes</b> , date of most recent immunisation			
DD/MM/YYYY			
If <b>No</b> , reason why			

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#### 24. Person's medical history

Please list any information about the person's health history, which has not been recorded in this booklet. Consider medical problems, surgery/operations, gynaecological, obstetric and psychiatric conditions

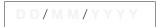
#### 25. Family medical history

Has anyone in the person's family (blood relation) had any of the following conditions?	Yes	No	Unsure/ don't know
Diabetes			
Psychiatric Disorders			
Heart Disease			
Epilepsy			
Breast Cancer			
Intellectual Disability			
Bowel Cancer			

#### 27. Person who completed Part 1 of this document

Name

#### Date completed



#### **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 – For the General Practitioner

Thank you for reviewing this person's health care.

Date of visit

DD/MM/YYYY

#### Review the history provided in the 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 / malnutrition
- 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 an intellectual disability are:

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

A health assessment for people with an 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
- screening areas often neglected
- other tests you feel are indicated, for example, a urine analysis.

#### Please record your findings on the following pages.

The last page of this book has syndrome specific information for you to keep.

Using this Comprehensive Health Assessment Program (CHAP) can improve the health of a person with intellectual disability. Several randomised controlled trials and observational studies have demonstrated its beneficial effect to detect new disease, 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

2.

Height	cm
Weight	kg
Blood Pressure	mmHg
Pulse Rate	BPM
If the person has a wei	ght / blood pressure problem, please specify action recommended or taken:
Weight control	
Hypertension	
ls waist measurement	necessary? Yes No
	ete comprehensive physical examination New findings
Cardiovascular	
Respiratory	
Musculo-skeletal	
Renal / urogenital	
Endocrine	
Gastrointestinal	
Nervous	
Psychiatric/ Behavioural	
Skin	

#### 3. Mental health

People with intellectual disability have very high rates of mental ill health which is often unrecognised. Is there any evidence of a comorbid psychiatric disorder?

## 4. Epilepsy

	If present	
	Has the person's seizure control been reviewed? Yes No	
	Does the person have an epilepsy management plan? Yes $\Box$ No $\Box$	
	Has the risk of SUDEP been minimised? Yes No	
	Has a referral been considered? Yes No	
5.	<b>Vision test</b> 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 & tympanometry	
	Has a hearing test been performed? Yes No	
	Test result	
	Right 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 First Recommend a CGH Microarray (Medicare indication: Intellectual disability)	
	Then Recommend testing for Fragile X, a urinary and plasma metabolic screen	
	And $\Box$ If no results, consider referring to a genetics clinic	
0	<i>,</i>	
8.	Sleep Does the person have sleep apnoea or sleep problems? Yes No	
•		
9.	Health of women and people with a cervix         Was a breast examination undertaken?       Yes         No	
	Is a mammogram required? Yes No	
	Was cervical screening undertaken/organised for the future (e.g. Supporting vaginal swab for HPV)? Yes No	
	If the person has symptoms which could be suspicious for	
	cervical cancer a co-test rather than a screening test should be ordered alongside other investigations as appropriate.	

10.	Health of men and people with a penis		
	Was the person checked for undescended testicles?	Yes	No
	Does this person need prostate screening?	Yes	No
11.	<b>Sexual and reproductive health</b> Recommend a review of their reproductive health, sexual activit	y and sexual	development.
12.	<b>Abuse and Trauma</b> Check for signs of physical, psychological or sexual abuse and	trauma.	
13.	Dental Health		
	Has the person been reviewed by a dentist in the last six months?	Yes	No
	Is there obvious dental pathology?	Yes	No
14.	<b>GIT and bladder function</b> 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
15.	Activity and lifestyle Inactivity and poor diet can lead to significant health problems a	and depressio	on
	Does the person have sufficient <b>exercise</b> ?	Yes	No
	Does the person have any <b>diet problems</b> / <b>abnormal eating behaviours</b> ?	Yes	No
	Is a referral needed for any of these?	Yes	No
16.	Health promotion and screening		
	Blood glucose tested?	Yes	No 🗌
	Lipid Screen tested?	Yes	No
	Thyroid function tested? Especially in people with Down syndrome	Yes	No
	Colorectal cancer assessment? Same risk factors as general population	Yes	No
	Osteoporosis risk assessed?	Yes	No

Vitamin D deficiency, poor diet, lack of exercise, hypogonadism and medication (including anti-psychotics & anti-convulsants) issues all appear to be common in some people with intellectual disability.

#### 17. Immunisations

Are the following immunisations indicated?

COVID-19	Yes	No
Tetanus, diphtheria & pertussis (dTpa)	Yes	No
Hepatitis A	Yes	No
Hepatitis B	Yes	No
Pneumococcus	Yes	No
Meningococcal	Yes	No
Measles, Mumps & Rubella	Yes	No
HPV cervical cancer	Yes	No
Herpes Zoster	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/

#### 18. Medication review

19.

Including prescription and non-prescription medications		
Have the person's medications been reviewed?	Yes	No
Have you communicated indications, side effects and interactions?	Yes	No
Future planning		
Have there been any <b>mobility</b> changes over time?	Yes	No
Has there been any <b>functional decline</b> over time?	Yes	No
Would you be surprised if the person were to die in the next 12 months?	Yes	No
If <b>No</b> , have you considered <b>advanced care</b> <b>planning and palliative care</b> , and discussed with the person, their family or carers?	Yes	No

If possible, please assist the 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 400,000+ 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 an 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)

N 1	~	
Name	Ot.	person

Address

## **ACTION PLAN**

Problem(s) identified	Action(s) to be Taken	<b>MEDICATION CHANGES</b> (Name of medication, how often, how much OR whether to cease)	Action(s) by, Arranged by:	By when (Date)	Actions completed & date

#### Comments or notes about the consultation

Details of **case conference** (including other professionals involved)

Completed by (please print) Signature

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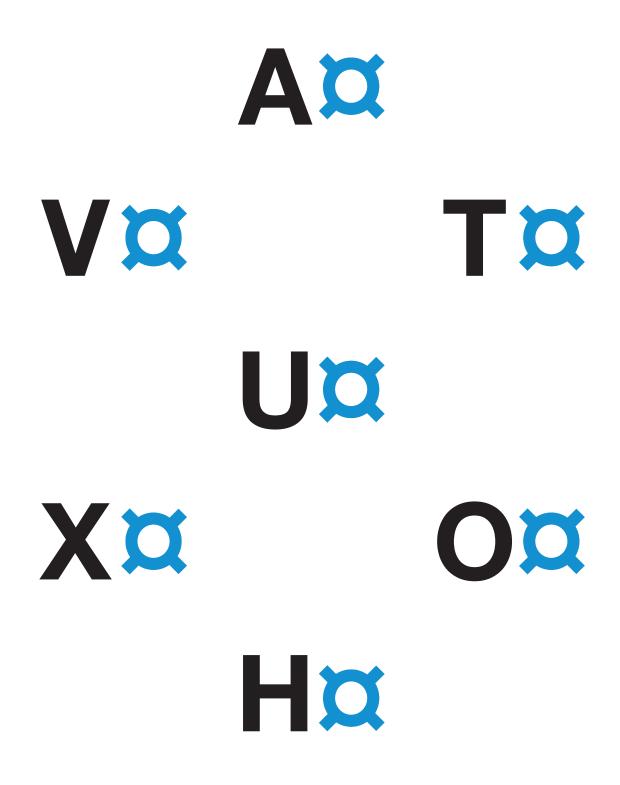
	Syndrome specific list for general practitioners					
SYSTEM	CEREBRAL PALSY 1:500	DOWN SYNDROME 1:700	PRADER-WILLI 1:25,000			
AUDIOVISUAL	Visual: Increased risk of cicatricial retinopathy of prematurity, cortical visual impairment, strabismus Hearing: Sensorineural hearing	Visual most common - Refractive errors (myopia, hyperopia, astigmatism), Strabismus, Nystagmus, Keratoconus, and cataracts Hearing: Otitis media affects 50 to 70% DS children, conductive and sensorineural hearing loss Annual assessments recommended	Strabismus Myopia			
ENDOCRINE		Thyroid dysfunction & diabetes (both hypo or hyperthyroidism) – Hypo is more common Annual TFT recommended Growth, short statue Obesity and diabetes Osteoporosis	Hypothalamic & pituitary dysfunction Growth hormone deficiency Adrenal insufficiency Hypothyroidism, Type 2 Diabetes, Hypogonadism, Osteoporosis Delayed puberty			
PSYCHIATRIC/ PSYCHOLOGICAL	Depression Not all have intellectual disability	Depression, anxiety, OCD Alzheimer's type dementia (Clinical onset uncommon before 40 years)	Hyperphagia Impulse control difficulties Self-injury Depression, anxiety, psychosis Not all have intellectual disability			
C.N.S.	Epilepsy	Epilepsy (Usually clonic/tonic)	Epilepsy			
CARDIOVASCULAR		Congenital Heart Defects (Common - in 40 to 50%)	Hypertension			
MUSCULO-SKELETAL	Contractures, scoliosis, joint dysfunction Managing pain, maintaining range of movement, muscle strength and general fitness Physiotherapy to monitor muscular function.	Joint laxity and hypotonia Predisposition to osteoarthritis Atlantoaxial dislocation, instability	Scoliosis Kyphosis Hypotonia			
ADDITIONAL	Genito-urinary problems Incontinence, Constipation Dental problems Recurrent aspiration Oesophagitis, Reflux +/- bleeding/ anaemia; Swallowing/eating difficulties - Speech disorders affect 40-50%; Abnormal function of oropharyngeal muscles Orthopaedic disorders; Osteopenia Pulmonary disease (caused by reflux & aspiration) Growth failure	Blood dyscrasias Childhood leukaemia – AML, ALL Sleep apnoea Increased risk of infections Coeliac disease Urological abnormalities Skin disorders, alopecia, eczema Gastrointestinal abnormalities	Infantile failure to thrive, then hyperphagia and severe obesity High tolerance to pain Decreased ability to vomit Sleep apnoea Undescended testes Dental Abnormalities Gastroesophageal reflux Skin picking			
INHERITANCE	75% prenatal causes 8-10% perinatal asphyxia	Most cases are sporadic; 4% due to translocation involving chromosome 21 or rarely, parental mosaicism	Atypical. Most cases are sporadic.			

SYSTEM	RETT	NOONAN	TUBEROUS SCLEROSIS	NEUROFIBRO MATOSIS
STSTEM	1:10,000 FEMALES	<1:10,000	1:6,000-17,000	Type 1 - 1:3,000
	1.10,000 FEMALES	<1.10,000	1.0,000-17,000	Type 2 - 1:30,000
AUDIOVISUAL	Refractory errors	Strabismus, refractive errors Vision/hearing impairments	Retinal tumours Eye rhabdomyomatas	Hearing impairment (Glioma affecting auditory nerve) Optic gliomas
ENDOCRINE				Various endocrine abnormalities
PSYCHIATRIC/ PSYCHOLOGICAL	Intellectual disability	Intellectual disability	Variable intellectual capacity Sleep problems Hyperactivity Aggressive behaviour Autism spectrum disorder Not all have intellectual disability	Increased risk of attention deficit disorders Not all have intellectual disability
C.N.S.	Epilepsy Vasomotor instability	Epilepsy	Cerebral astrocytomas Epilepsy	Variable clinical phenomena depending on site of the tumours Epilepsy
CARDIOVASCULAR	Prolonged QT interval	Pulmonary Valvular Stenosis ASD, VSD, PDA	Rhabdomyomatas Hypertension	Rarely structural cardiac complications Increase in blood pressure with age
MUSCULO- SKELETAL	Osteopenia Fractures Scoliosis Gate abnormality	Scoliosis Talipes equinovarus Pectus carinatum/ excavatum	Bone Rhabdomyomata	Skeletal abnormalities esp. Kyphoscoliosis
ADDITIONAL	Hyperventilation Apnoea Reflux Feeding difficulties Growth failure Partial or complete loss of acquired spoken language Partial or complete loss of purposeful hand skills Impaired sleep pattern	Abnormal clotting factors, Platelet dysfunction Undescended testes, Deficient spermatogenesis Lymphoedenoma Hepatosplenomegaly Cubitus valgus, Hand abnormalities Characteristic facial features Short stature (80%)	Kidney & lung hamartomata Polycystic kidneys Liver Rhabdomyomata Dental abnormalities Skin lesions Subependymal nodules	Variable clinical phenomena depends on neurofibroma location Tumours susceptible to malignant change Cafe au lait spots Axillary and inguinal freckling Usually back pain & headaches Phaeochromocytoma & renal artery stenosis rare
INHERITANCE	Usually sporadic. X linked	Autosomal dominant, may be sporadic	Autosomal dominant	Autosomal dominant

	Syn	drome specific list for gener	al practitioners	
SYSTEM	FRAGILE X 1:4,000 to 7,000 <sup>1</sup>	PHENYLKETONURIA 1:13,500-1:19,000	ANGELMANN SYNDROME 1:10,000-52,000	WILLIAMS <1:20,000
AUDIOVISUAL	Visual Impairment – Multifactorial, Strabismus Hearing Impairment - Recurrent ear infections		Glaucoma Strabismus is possible	Hyperacusis Strabismus
ENDOCRINE	Primary ovarian failure in carriers			Diabetes Hypercalcaemia Hypothyroidism
PSYCHIATRIC/ PSYCHOLOGICAL	Attention deficit/ hyperactivity Social difficulties Anxiety & Autism Spectrum Disorders Not all have intellectual disability	Phobic anxiety Social difficulties Excessive sadness, fear & anxiety; sense of isolation & poor self-image Not all have intellectual disability	Easily excitable Hyperactive Intellectual disability	Attention deficit problems in childhooc Not all have intellectual disability
C.N.S.	Epilepsy (Usually clonic/tonic, complex partial)	Epilepsy Hyperactivity Tremor & pyramidal tract signs Extrapyramidal syndromes	Epilepsy	Perceptual & motor function reduced
CARDIOVASCULAR	Aortic dilatation, Mitral Valve prolapse (related to connective tissue dysplasia)			Cardiac abnormalities Hypertension, CVAs Chronic hemiparesis
MUSCULO- SKELETAL	Connective tissue dysplasia Scoliosis Congenital hip dislocation	Osteopenia	Joint contractures & scoliosis (in adults)	Joint contractures Scoliosis Hypotonia
ADDITIONAL	FXTAS in carriers Herniae (CT related) Abnormalities of speech & language Avoidant behaviour Delayed language development Obesity & hyperphagia	Eczema Low phenylalanine diet provides considerable benefit	Speech impairment, Movement & balance disorder; Characteristic EEG changes; Gastrointestinal problems; Orthopaedic problems; Sleep problems; Obesity; Microcephaly	Renal abnormalities Short stature Characteristic facial features (elfin) Gastrointestinal problems
INHERITANCE	X linked	Autosomal recessive	Variety of genetic mechanisms on Chromosome 15	Microdeletion on chromosome 7

Adapted from an original unpublished version by Michael Kerr and Glyn

<sup>1</sup> Permutation prevalence – 1 in 130-250 females; 1 in 250-810 males (see: <a href="http://www.irdrjournal.com/files/IRDR\_2014Vol3No4\_pp100\_182.pdf">http://www.irdrjournal.com/files/IRDR\_2014Vol3No4\_pp100\_182.pdf</a>)



If the 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 May 2023

