



**Human Services**  
Ageing, Disability & Home Care

# C · H · A · P

COMPREHENSIVE · HEALTH  
ASSESSMENT · PROGRAM

Version 8

**UNDER LICENSE FROM:  
Queensland Centre for Intellectual and  
Developmental Disability**



**THE UNIVERSITY  
OF QUEENSLAND**



PERSON (with intellectual disability) \_\_\_\_\_

FAMILY MEMBER / SUPPORT STAFF \_\_\_\_\_

GENERAL PRACTITIONER \_\_\_\_\_

## STEPS TO FOLLOW:

1. Person, family member and/or support staff is to fill in the first part of this book for the person. It is OK to get information from records, family members and staff.
2. Make a long appointment for the person with his/her usual GP.
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 the second part while examining the person.
5. It is important that the *Action Plan* is filled in at the end of the consultation. One copy of the *Action Plan* is to be kept with the person's personal record.
6. Medicare supports this assessment. Ask the GP.

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Queensland Centre for Intellectual and Developmental Disability,

The University of Queensland



Please tick in 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.

1). BREATHING SYSTEM	YES	NO	Unsure/ don't know
Does the person <b>cough</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person cough up <b>blood</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person cough up stuff/mucous/ <b>sputum</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person get short of <b>breath</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person <b>wheeze</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

2). HEART SYSTEM	YES	NO	Unsure/ don't know
Does the person have <b>chest pain</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person's heart "race"/ <b>beat quickly</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Do the person's <b>ankles swell</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person get <b>short of breath</b> while lying in bed?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person get <b>blue skin</b> (e.g. fingers/lips/toes)?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

3). MUSCLES & JOINTS	YES	NO	Unsure/ don't know
Does the person have joint pain or <b>back pain</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person have <b>muscle pain</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**4). STOMACH & BOWEL SYSTEM****YES****NO****Unsure/  
don't know**

Has the person <b>lost weight</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person have <b>trouble swallowing</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person <b>regurgitate/vomit</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person get " <b>heart burn</b> "?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person have <b>diarrhoea</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person have <b>black bowel motions</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person get <b>constipated</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person lose control of <b>bowel movements</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person have <b>abdominal/stomach pain</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

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**5). URINARY SYSTEM****YES****NO****Unsure  
don't know**

Does the person have <b>pain</b> when passing <b>urine</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person have <b>blood</b> in the <b>urine</b> ?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person lose <b>control passing urine</b> /incontinence?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Does the person urinate <b>a lot</b> /more than usual?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

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**6). NERVOUS SYSTEM****YES****NO****Unsure/  
don't know****(Please note that epileptic episodes are not included here)**

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

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## 7). MEDICATIONS

It is important for the doctor to know ALL medication.

List the medication/s prescribed by any doctor.

Name	How often and how much is taken?

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

Name	How often and how much is taken?



## 8). ALLERGIES

Please list any medications the person is allergic to:

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9). CAUSE OF INTELLECTUAL DISABILITY	YES	NO	Unsure/ don't know
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Is the cause of intellectual disability known?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
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If **YES** → Please state cause:

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**10). EPILEPSY****YES****NO****Unsure/  
don't know**

During the person's life, has he or she had epileptic seizures/ fits?

If **NO** → Please go to **Question 11**If **YES** → Please list the **type** and **number** of seizures during the last year.

Type of Seizures	Number of Seizures

Which doctor treats the epileptic seizures/fits?

**(Please tick one)**

GP

Neurologist

Other:

\_\_\_\_\_

When did this doctor last review the person's epilepsy?

(month and year if known)

\_\_\_/\_\_\_/\_\_\_

Unknown

Since this review, have the seizures:

**(Please tick one)**

Become worse

Remained the same

Improved





**WOMEN'S HEALTH (cont'd)**

**Women with intellectual disability have the same reason for needing a Pap Smear test as women in the general population. A test every two years is recommended for women (between 18 and 70 years) who have ever been sexually active.**

	YES	NO	Unsure/ don't know
Has the person had a Pap smear?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
If <b>YES</b> → When was the last test: ___/___/___			
If <b>NO</b> → Does the person need a Pap smear?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
If a Pap smear is needed but has not happened, please indicate why:			
Distress	<input type="checkbox"/>		
Pap smear planned	<input type="checkbox"/>		
Desensitisation/preparation planned	<input type="checkbox"/>		
Desensitisation/preparation in progress	<input type="checkbox"/>		
Other:	<input type="checkbox"/>		

**A mammogram should be arranged every 2 years for women over 50 years of age, or women with a family history of breast cancer.**

	YES	NO	Unsure/ don't know
Does the person check her own breasts monthly?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
If <b>NO</b> → Are the person's breasts checked by a GP?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Has the person ever had a mammogram?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
If <b>YES</b> → When was the last mammogram? ___/___/___			
If a mammogram is needed but has not happened, please indicate why:			
Distress	<input type="checkbox"/>		
Mammogram planned	<input type="checkbox"/>		
Desensitisation/preparation planned	<input type="checkbox"/>		
Desensitisation/preparation in progress	<input type="checkbox"/>		
Other:	<input type="checkbox"/>		

**FOR WOMEN, PLEASE GO TO QUESTION 14 ON THE NEXT PAGE**



**13). MEN'S HEALTH**

**YES**

**NO**

**Unsure/  
don't know**

Does the person have a discharge from his penis?

Does the person have any sores or scars on his penis?

**14). PROBLEM BEHAVIOURS**

**YES**

**NO**

**Unsure/  
don't know**

Does the person have any problem behaviours?

If **YES** → Please describe the behaviours and any help the person receives for these behaviours:

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**15). MENTAL HEALTH**

**YES**

**NO**

**Unsure/  
don't know**

Does the person have a psychiatric illness?

If **YES** → Please describe the psychiatric illness and any help the person receives for the psychiatric illness:

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**16). VISION**

**YES**

**NO**

**Unsure/  
don't know**

Does the person have a known problem with vision?  
If **YES** → Please describe the problem:

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If **NO**

**YES**

**NO**

**Unsure/  
don't know**

Do you suspect the person may have a problem with vision?

Has the person ever been prescribed glasses?

Does the person usually wear the glasses?

When was the last vision test?

(month and year if known)    \_\_\_/\_\_\_

Unknown

Never

Who performed this test?

Eye doctor / ophthalmologist

Optometrist

GP

Test while at school

Unknown

Result of last vision test:

Normal

Unknown

Abnormality found → Please describe:

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**17). HEARING**

**YES**

**NO**

**Unsure/  
don't know**

Does the person have a problem with hearing?

If **YES** Please describe the problem:

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If **NO**

**YES**

**NO**

**Unsure/  
don't know**

Do you suspect the person has a hearing problem?

When was the person's last hearing test?

(month and year if known) \_\_\_/\_\_\_

Unknown

Never

Who performed this test?

Audiologist

GP

Other: \_\_\_\_\_

Result of last hearing test:

Normal

Unknown

**YES**

**NO**

**Unsure/  
don't know**

Has the person been prescribed a hearing aid?

Does the person usually wear the hearing aid?



**18). HEALTH PROMOTION & SCREENING**

a). **Dental:** Date of last review: \_\_\_/\_\_\_/\_\_\_ Unknown

b). **Blood pressure:** Date of last check: \_\_\_/\_\_\_/\_\_\_ Unknown

c). **Cigarettes** smoked (per day):

d). **Alcohol** (Standard drinks per week):

e). **Thyroid function test (TFT):** If the person has Down syndrome, when was the most recent test?

Date: \_\_\_/\_\_\_/\_\_\_ Unknown

f). **Vitamin D test:** Date of last review: \_\_\_/\_\_\_/\_\_\_ Unknown

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

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**19). ACTIVITY & LIFESTYLE**

a) Have there been any mobility changes over time? Yes  No

b) Does the person have sufficient exercise? Yes  No

c) Does the person have any diet problems or abnormal eating behaviours?  
Yes  No

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**20). IMMUNISATIONS**

**YES NO Unsure/don't know**

a). **Tetanus/Diphtheria/Pertussis**

If **YES** → Date of most recent immunisation: \_\_\_/\_\_\_/\_\_\_

If **NO** → Reason why: \_\_\_\_\_

b). **Hepatitis A & B (If required- check with the GP)**

If **YES** → Date of most recent immunisation: \_\_\_/\_\_\_/\_\_\_

If **NO** → Reason why: \_\_\_\_\_

c). **Influenza (If required- check with the GP)**

If **YES** → Date of most recent immunisation: \_\_\_/\_\_\_/\_\_\_

If **NO** → Reason why: \_\_\_\_\_

d). **Pneumococcus (If required - check with the GP)**

If **YES** → Date of most recent immunisation: \_\_\_/\_\_\_/\_\_\_

If **NO** → Reason why: \_\_\_\_\_

e). **Measles, Mumps and Rubella (Check with the GP)**

If **YES** → Date of most recent immunisation: \_\_\_/\_\_\_/\_\_\_

If **NO** → Reason why: \_\_\_\_\_

f). **Cervical Cancer – for women (Check with the GP)**

If **YES** → Date of most recent immunisation: \_\_\_/\_\_\_/\_\_\_

If **NO** → Reason why: \_\_\_\_\_

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**21). 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:

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**22). 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	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Psychiatric Disorders	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Heart Disease	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Epilepsy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Breast Cancer	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Intellectual Disability	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**23). SUMMARY OF HEALTH CONCERNS**

List the concerns that you have about the person's health.

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**For the GENERAL PRACTITIONER**

**Thank you for reviewing this person's health care**

Date of visit \_\_\_/\_\_\_/\_\_\_\_\_

- A. Please review the history provided in the First section.
- B. The following list shows commonly neglected areas of health in this population:
- hearing and vision impairment +/- unrecognised pathology
  - incomplete immunisation schedules
  - health screens - BP, skin, breast, Pap smear
  - obesity / malnutrition
  - over-use and inadequate review of tranquillisers/anti-convulsants
  - unrecognised constipation
  - unrecognised reflux oesophagitis / H.pylori infection / dysphagia
  - psychiatric assessment / management
  - epilepsy assessment / management
  - unrecognised pain or infections
  - poor dental care
  - unrecognised osteoporosis
  - undescended testes/ hypogonadism
  - Vitamin D deficiency
  - information about menstrual management
  - information about human relations.

**There have been changes to the MBS. There are now four Medicare Benefits Schedule items for health assessments based on how long the assessment takes (items 701, 703, 705 & 707).**

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

Full physical examination  
Screening areas often neglected  
Urine analysis  
Other tests you feel are indicated

Please record your findings on the following pages

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



1). **Height:** \_\_\_ cm    **Weight:** \_\_\_ kg    **Blood Pressure:** \_\_\_ mmHg    **Pulse Rate** \_\_\_

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

Weight control: \_\_\_\_\_

Hypertension: \_\_\_\_\_

2). **Systems Check**

Please perform a complete comprehensive physical examination

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### NEW FINDINGS

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CARDIOVASCULAR

RESPIRATORY

MUSCULO-SKELETAL

RENAL / UROGENITAL

ENDOCRINE

GASTROINTESTINAL

NERVOUS

PSYCHIATRIC/  
BEHAVIOURAL

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3). **Mental Health**

Is there any evidence of an underlying psychiatric disorder?

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**4). Epilepsy (if present)**Has the person's seizure control been reviewed? **Yes**  **No** Has a referral been considered? **Yes**  **No** **5). Vision test** (A special "Reading Card" is on the back of this book to aid testing)Has the person's vision been tested? **Yes**  **No** Test result: **Both eyes** \_\_\_\_\_ **R)** \_\_\_\_\_ **L)** \_\_\_\_\_ 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 an otoscopy been performed? **Yes**  **No** Test result: \_\_\_\_\_ **R)** \_\_\_\_\_ **L)** \_\_\_\_\_ Unable to test Uncertain

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

**7). Aetiology****If there is no definitive diagnosis:**

Recommend testing for karyotype and Fragile X, and doing a urinary and plasma metabolic screen

AND

Recommend the person is referred to a genetics clinic every five years

**8). Women's Health**Was a breast examination undertaken? Yes  No Was a Pap smear test taken/organised for the future? Yes  No **9). Men's Health**Was the person checked for undescended testicles? Yes  No Does this person need prostate screening? Yes  No **10). Reproductive health**

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

**11). Abuse**

Check for signs of physical, psychological or sexual abuse

**12). Dental Health**Has the person been reviewed by a dentist in the last six months? Yes  No Is there obvious dental pathology? Yes  No **13). Dysphagia or gastro-oesophageal disease** (especially for people with cerebral palsy)Has the person been assessed? Yes  No **14). Bowel and bladder function**Does the person experience incontinence? Yes  No Does the person have chronic constipation? Yes  No **15). Activity and Lifestyle**Have there been any **mobility** changes over time? Yes  No 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

**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

**Osteoporosis risk assessed?** Yes  No

(Vitamin D deficiency, poor diet, lack of exercise, hypogonadism and medication issues appear to be common in some people with intellectual disability.)

**Colorectal cancer assessment?** (Same risk factors as general population) Yes  No

**17). Immunisations -** Are the following immunisations indicated?

Tetanus, diphtheria & pertussis (dTpa) Yes  No

Hepatitis A & B Yes  No

Influenza Yes  No

Pneumococcus Yes  No

Measles, Mumps & Rubella Yes  No

HPV cervical cancer Yes  No

Influenza and Pneumococcus may be indicated for people with Down syndrome and others at risk of infectious disease.  
For more details, please refer to *The Australian Immunisation Handbook*.  
<http://www.immunise.health.gov.au/>

**18). Medication Review** (including prescription and non-prescription medications)

Have the person's medications been reviewed? Yes  No

Consider indications, side effects and interactions

**We recommend that you 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 needs to be returned to the care provider who came to this consultation.**  
*The last 3 pages of this book are for you to tear out and keep as your reference.*

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

GP-4



Name of Person: \_\_\_\_\_ Address: \_\_\_\_\_

## ACTION PLAN

Problem(s) Identified	Action(s) to be Taken	MEDICATION CHANGES (Name of medication, how often, how much OR whether to cease)	Action(s) by, Arranged by:	By when (Date)

Comments or notes about the consultation:

\_\_\_\_\_  
\_\_\_\_\_

Details of CASE CONFERENCE (including other professionals involved):

\_\_\_\_\_  
\_\_\_\_\_

Completed by: (please print): \_\_\_\_\_ Signature: \_\_\_\_\_



Name of Person: \_\_\_\_\_ Address: \_\_\_\_\_

## ACTION PLAN

Problem(s) Identified	Action(s) to be Taken	MEDICATION CHANGES (Name of medication, how often, how much OR whether to cease)	Action(s) by, Arranged by:	By when (Date)

Comments or notes about the consultation:

\_\_\_\_\_  
\_\_\_\_\_

Details of CASE CONFERENCE (including other professionals involved):

\_\_\_\_\_  
\_\_\_\_\_

Completed by: (please print): \_\_\_\_\_ Signature: \_\_\_\_\_



SYNDROME SPECIFIC LIST FOR GENERAL PRACTITIONERS

	<b>CEREBRAL PALSY 1:500</b>	<b>DOWN SYNDROME 1:700</b>	<b>PRADER-WILLI 1:10 000-25 000</b>	<b>FRAGILE X 1:6 000</b>	<b>PHENYL/ KETONURIA 1:10 000-1:20 000</b>
<b>AUDIOVISUAL</b>	Visual Impairment Hearing Impairment	Visual impairment (multifactorial), Cataracts Hearing impairment (multifactorial)  (Annual assessments recommended)	Strabismus  Myopia	Visual Impairment (Multifactorial)  Hearing Impairment Recurrent ear infections	
<b>ENDOCRINE</b>		Hypothyroidism (Annual TFT recommended)	NIDDM (secondary to obesity) Hypogonadism Delayed puberty		
<b>PSYCHIATRIC/ PSYCHOLOGICAL</b>	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
<b>C.N.S.</b>	Epilepsy	Epilepsy (Usually clonic/tonic)		Epilepsy (Usually clonic/tonic, complex partial)	Epilepsy Hyperactivity Tremor & pyramidal tract signs Extrapyramidal syndromes
<b>CARDIOVASCULAR</b>		Congenital Heart Defects (Common - in 40 to 50%)		Aortic dilatation, Mitral Valve prolapse (related to connective tissue dysplasia)	
<b>OTHER</b>	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
<b>INHERITANCE</b>		Most cases are sporadic; 4% due to translocation involving chromosome 21 or rarely, parental mosaicism	Atypical. Most cases are sporadic.	X linked	Autosomal recessive



**Comprehensive Health Assessment Program (CHAP) © 2010**  
**SYNDROME SPECIFIC LIST FOR GENERAL PRACTITIONERS**



	<b>ANGELMANN SYNDROME</b> <b>&lt;1:10 000</b>	<b>WILLIAMS</b> <b>?&lt;1:20 000</b>	<b>RETT</b> <b>1;14 000</b> <b>FEMALES</b>	<b>NOONAN</b> <b>&lt;1:10 000</b>	<b>TUBEROUS SCLEROSIS</b> <b>1:6 000-17 000</b>	<b>NEUROFIBRO MATOSIS</b> <b>1:3 000</b>
<b>AUDIOVISUAL</b>	Glaucoma	Hyperacusis Strabismus	Refractory errors	Strabismus, refractive errors Vision/hearing impairments	Retinal tumours Eye rhabdomyomas	Hearing impairment (Glioma affecting auditory nerve)
<b>ENDOCRINE</b>						Various endocrine abnormalities
<b>PSYCHIATRIC/ PSYCHOLOGICAL</b>	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
<b>C.N.S.</b>	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
<b>CARDIOVASCULAR</b>		Cardiac abnormalities Hypertension, CVAs Chronic hemiparesis	Prolonged QT interval	Pulmonary Valvular Stenosis ASD, VSD, PDA	Rhabdomyomas Hypertension	
<b>MUSCULAR/ SKELETAL</b>	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
<b>OTHER</b>	Speech impairment Movement & balance disorder Characteristic EEG changes	Renal abnormalities	Hyperventilation Apnoea Reflux Feeding difficulties Growth failure	Abnormal clotting factors, Platelet dysfunction Undescended testes, Deficient spermatogenesis Lymphoedema Hepatosplenomegaly Cubitus valgus, Hand abnormalities	Kidney & 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
<b>INHERITANCE</b>	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

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