

## **Syndrome specific check at AHC**

### **Down's syndrome**

Website for health care professionals: <https://www.dsmig.org.uk/>

With guidance for surveillance: <https://www.dsmig.org.uk/information-resources/guidance-for-essential-medical-surveillance/>

### **Fragile x-syndrome**

Website mainly aimed at families/carers : <https://www.fragilex.org.uk/>

### **Rett Syndrome**

Website: <https://www.rettuk.org/>

PDF Rett best practice guidance: <https://www.rettuk.org/wp-content/uploads/2015/03/Rett-UK-Management-Care-Guidelines.pdf>

Dyscerne <https://dyscerne.org/dysc/Guidelines> has detailed clinical management guidelines for four dysmorphic conditions.

- **Angelman syndrome**
- **Kabuki syndrome**
- **Noonan syndrome**
- **Williams syndrome**

## **Syndrome specific check old RCGP AHC template *(useful links added in brackets)***

### **Adult Down's Syndrome Specific Annual Health Check list**

<b>HISTORY</b>	<ul style="list-style-type: none"><li>➤ Because of the high prevalence of hearing impairment check the person can hear you at the start of the health check.</li></ul> <p>As with all people with LD focus on</p> <ul style="list-style-type: none"><li>• Assessment of feeding, bowel and bladder function</li><li>• Assessment of behavioural disturbance</li><li>• Assessment of vision and hearing</li></ul> <ul style="list-style-type: none"><li>➤ Dementia: Monitor for any loss of independence in living skills, behavioural changes and/or mental health problems.</li></ul>
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	<p>Look for symptoms of dementia (decline in function, memory loss, ataxia, seizures or urinary and/or faecal incontinence).</p> <p>Check that people with a diagnosis of Alzheimer’s disease have had depression, hypothyroidism, and deafness excluded.</p> <ul style="list-style-type: none"> <li>➤ Ask about sleep apnoea which may due to a hypoplastic Pharynx or nasal congestion.</li> <li>➤ Ask about hot flushes and menopausal symptoms in women over 40 as they have an earlier onset of menopause compared to women in the general population at 44 years of age. Women with Down Syndrome with an early onset of menopause also appear to suffer from dementia at an early age and die younger</li> </ul>
	<p><b>Examination</b></p>
audio-visual	<p>Ophthalmic Problems (cataract, glaucoma, keratoconus and refractive errors).</p> <ul style="list-style-type: none"> <li>➤ Full assessment by optician/optometrist every 2 years</li> <li>➤ If examination difficult, refer to specialist optician or ophthalmologist for assessment.</li> </ul> <p>(SeeAbility <a href="http://www.seeability.org">www.seeability.org</a> has a tool to find a local optometrist for patients with learning disability <a href="https://www.seeability.org/optometrists">https://www.seeability.org/optometrists</a> )</p> <p>Audiological problems (hearing impairment and deafness)</p> <ul style="list-style-type: none"> <li>➤ Otoscopy (Gentle examination as short ext. auditory canals)</li> <li>➤ Audiological Assessment every 2 years (including auditory thresholds, impedance testing)</li> </ul> <p>Well over 50% of people with Down’s syndrome have significant hearing impairment, which can range from mild to profound. Sensorineural and/ or conductive loss may be present at any age. If undetected it is likely to be a significant cause of preventable secondary handicap. The main cause of conductive loss is persistent otitis media with effusion (OME, glue ear).</p>
dental	<ul style="list-style-type: none"> <li>➤ Annual Dental Review as periodontal disease is common.</li> <li>➤ Look for Signs of oesophageal reflux</li> </ul>
endocrine	<p>There is an increased prevalence of hypothyroidism at all ages, rising with age with a small increase in hyperthyroidism.</p>

	<ul style="list-style-type: none"> <li>➤ Thyroid Function blood tests (TFTs), including thyroid antibodies, at least every 2 years,</li> </ul> <p>Perform TFTs more often if</p> <ul style="list-style-type: none"> <li>• accelerated weight gain</li> <li>• unwell</li> <li>• possible diagnosis of depression or dementia.</li> </ul> <p>Type I diabetes is also relatively more common (2%).</p>
Psychiatric/ psychological	<p>Alzheimer's type dementia (clinical onset uncommon before 40 years), which often presents as deterioration in self help skills or behaviour change.</p> <p>Need to exclude depression, thyroid disorder and hearing impairment.</p> <p>Depression is common in older adults, often as a result of bereavement and/or changes in living situation.</p>
cardiovascular	<p>Examine for adult onset mitral valve prolapse and aortic regurgitation.</p> <ul style="list-style-type: none"> <li>➤ Auscultation – particularly if imminent dental procedure</li> <li>➤ A single ECHO should be performed in adult life</li> <li>➤ Adults with a pre-existing structural abnormality should be informed of current prophylactic antibiotic protocols</li> </ul>
respiratory	<p>Examine nose, oral cavity and lungs</p> <ul style="list-style-type: none"> <li>➤ Blocked nasal passages</li> <li>➤ Lower airway disease</li> </ul>
Coeliac disease	<p>Screen clinically by history and examination annually.</p> <p>Testing in those with suspicious symptoms or signs, including</p> <ul style="list-style-type: none"> <li>➤ Disordered bowel function tending to diarrhoea or to new onset constipation</li> <li>➤ Abdominal distension</li> <li>➤ General unhappiness and misery</li> <li>➤ Arthritis</li> <li>➤ Rash suggesting dermatitis herpetiformis</li> <li>➤ test all those with existing thyroid disease, diabetes or anaemia.</li> </ul>
MSK	<p>Atlanto axial instability</p> <p>Most cases have been described in children with longitudinal studies of children and adults show a high degree of stability both clinically and radiologically.</p> <p>Routine Cervical -spine X-ray not recommended.</p> <p>It can present as acute or chronic cord compression:</p> <ul style="list-style-type: none"> <li>➤ Neck Pain</li> <li>➤ Reduced range of neck movement</li> <li>➤ torticollis</li> </ul>

	<ul style="list-style-type: none"> <li>➤ Unsteadiness</li> <li>➤ Deterioration in bladder / bowel control</li> </ul> <p>( more details here <a href="https://www.dsmig.org.uk/wp-content/uploads/2015/09/CSI-revision-final-2012.pdf">https://www.dsmig.org.uk/wp-content/uploads/2015/09/CSI-revision-final-2012.pdf</a> )</p> <p>Osteoporosis: Women with Down’s Syndrome reach the menopause approximately 6 years earlier than the general population and are more susceptible to osteoporosis particularly if they are inactive.</p>
other	<ul style="list-style-type: none"> <li>➤ Blood Dyscrasias</li> <li>➤ skin disorders</li> <li>➤ obesity- check weight changes</li> <li>➤ increased susceptibility to infection disease</li> </ul>

### Adult Fragile X Syndrome Specific Annual Health Check list

HISTORY	<p>As the most common cause of inherited learning difficulty, they have a normal life expectancy and generally have less severe medical complications.</p> <p>It affects males more than females and has a characteristic physical appearance:</p> <ul style="list-style-type: none"> <li>• long face</li> <li>• large jaw</li> <li>• prominent ears</li> <li>• enlarged testicles (post puberty)</li> <li>•</li> </ul> <p>As with all people with LD focus on</p> <ul style="list-style-type: none"> <li>➤ Assessment of feeding, bowel and bladder function</li> <li>➤ Assessment of behavioural disturbance</li> <li>➤ Assessment of vision and hearing</li> <li>➤</li> </ul> <p>Ask about anxiety (often highly anxious and overwhelmed), hyperactivity, autistic type features (such as hand flapping, biting, poor eye contact and shyness), ataxia, seizures and any joint dislocations (particularly patella and shoulder).</p> <p>In women ask about hot flushes as premature ovarian failure can occur before 30.</p>
EXAMINATION Audio-visual	<p>Eye problems can include squint (strabismus), long sightedness and visual perceptual problems.</p> <p>Eyelids tend to puffiness.</p> <ul style="list-style-type: none"> <li>➤ Full assessment by optician/optometrist every 2 years</li> </ul>

	<ul style="list-style-type: none"> <li>➤ Children with fragile X are prone to recurrent Otitis media.</li> <li>➤ Assessment including using whisper test and refer if concerns</li> </ul>
Abdominal	<ul style="list-style-type: none"> <li>➤ Examine the abdomen and inguinal areas as hernias are more common due connective tissue disorder.</li> </ul> <p>Men develop enlarged testicles (macroorchidism) after puberty, but this does not seem to pose any medical problems.</p>
cardiovascular	<p>Examine for adult onset mitral valve prolapse and aortic regurgitation.</p> <ul style="list-style-type: none"> <li>➤ Auscultation annually</li> </ul>
CNS	<p>About one in four people with fragile X have epilepsy which can be System generalised or focal (grand mal, petit mal or absences, or complex partial seizures). Seizures usually begin in childhood or adolescence and are not frequent, often being outgrown before adulthood.</p>
MSK	<p>Problems with connective tissue can lead to flat feet and low muscle tone. The joints are often extremely flexible and may dislocate.</p> <ul style="list-style-type: none"> <li>➤ Assessment spine for scoliosis</li> </ul>

### **Adult Rett's Syndrome Specific Annual Health Check list**

HISTORY	<p>As with all people with LD focus on</p> <ul style="list-style-type: none"> <li>➤ Assessment of feeding</li> <li>➤ bowel and bladder function</li> <li>➤ Assessment of behavioural disturbance</li> <li>➤ Assessment of vision and hearing</li> </ul> <p>Dr Alison Kerr has written a Clinical Check list for Retts syndrome (<a href="http://www.rettuk.org">www.rettuk.org</a>) and advises the following:</p> <ul style="list-style-type: none"> <li>• Communication is vital for the wellbeing of the individual. Assess capacities to understand speech, signs, symbols and written words and to find reliable means of expressive communication. Face to face communication is good and is usually more important than mechanical aids. One to one musical interaction is particularly valuable, encouraging choice, self expression, shared pleasure and control of the hands and voice Breathing rhythm is usually normal asleep and abnormal on alerting.</li> <li>• Apneustic breathing (prolonged inspiration) occurs mainly in younger and Valsalva breathing in older women. Shallow</li> </ul>
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	<p>breathing, breath holding and central apnoeas may lead to severe hypoxia.</p> <ul style="list-style-type: none"> <li>• Non-epileptic vacant spells are more frequent than epileptic seizures in Rett and are due to reduced brain stem cardio-respiratory control. This may lead to episodes of loss of consciousness, which may be difficult to differentiate from epilepsy and may require concurrent monitoring of central autonomic function with electroencephalography. Vagal tone and baroreflex sensitivity are usually reduced.</li> <li>• Dystonic spasms are common. Gentle massage may be more effective than medication.</li> <li>• Osteoporosis has been reported in Rett, even in active people. A balance must be found between providing active movement, which is essential for health and adequate protection from trauma.</li> <li>• Periodic unexplained agitation, laughing or crying is common and may be associated with the poor central parasympathetic restraint. It is helped by a quiet and relaxed atmosphere. Agitation is also the means to express any pain, irritation, discomfort, distress, anger, frustration or boredom and such causes must be carefully excluded. Sedatives and antipsychotics should be avoided. Short term use of a serotonin reuptake inhibitor may be helpful in extreme cases.</li> <li>• Sleep disorder: may include failure to go to sleep, night time waking and day time sleeping. Active days help to ensure quiet nights and bed time routines are helpful. The individual should sleep alone with a 'baby alarm' if necessary and intervention should be minimal. The room should be warm and safe to move about in without risk of injury. Melatonin may help to establish a routine.</li> </ul>
EXAMINATION dental	Check Teeth for grinding (bruxism) and ensure regular tooth cleaning and visits to the dentist.
Abdominal	<p>Poor feeding may be due to postural problems and reflux is common.</p> <ul style="list-style-type: none"> <li>➤ Examine the abdomen for constipation and abdominal distension due to aerophagy which commonly accompanies the abnormal breathing. Very severe cases may be helped by per-cutaneous gastrostomy.</li> </ul>
CNS	Epilepsy is present in about 50% and may remit. Generalised motor or System partial seizures respond to medication according to type. Since the electroencephalogram may be epileptogenic

	<p>without clinical epilepsy, video during prolonged recording may be necessary to distinguish epilepsy from non- epileptic vacant spells (see above).</p> <ul style="list-style-type: none"> <li>• Check seizure control and medication at each visit, Expect to wean off anticonvulsants if seizures become infrequent.</li> </ul> <p>Hand stereotypy is involuntary &amp; increased by alerting. It can be ignored unless injury occurs, when a light elbow splint may be used to prevent injury with minimal interference.</p> <p>Task performance may improve with one hand gently held (only during the task)</p>
Cardio-vascular	<p>Examine the feet and legs for poor blood circulation to the lower legs and feet (vasomotor disturbances) . Consider sympathectomy if severe.</p>
MSK	<ul style="list-style-type: none"> <li>• Review posture and joint position.</li> </ul> <p>Posture and joint position are likely to deteriorate due to initial hypotonia and later hypertonia.</p> <p>Large joints of shoulders, hips, knees and ankles are at risk of permanent flexion or extension of affected joints in fixed postures (joint contractures).</p> <p>Scoliosis is common with deterioration of back position during growth spurts. Ensure the person is receiving postural care and refer to orthopaedic surgeons for more severe or progressing curves.</p> <p>Hand skills are usually poor (dyspraxic) but improve given opportunity and encouragement. Gentle massage of the hands just before a task may encourage use eg holding mug or spoon within the adult's hand in feeding.</p>

### **Adult Williams Syndrome Specific Annual Health Check list**

HISTORY	<p>Williams syndrome is a sporadic genetic disorder due to deletion of a small part of chromosome 7.</p> <ul style="list-style-type: none"> <li>• Features may include a distinctive facial appearance, congenital heart defects and high levels of calcium in infancy.</li> <li>• Early feeding problems are common and development is delayed.</li> </ul>
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	<ul style="list-style-type: none"> <li>• People with WS have sociable personalities, characteristic behavioural traits and variable degrees of learning disability.</li> </ul> <p>As with all people with LD focus on</p> <ul style="list-style-type: none"> <li>➤ Assessment of feeding, bowel and bladder function</li> <li>➤ Assessment of behavioural disturbance</li> <li>➤ Assessment of vision and hearing</li> <li>➤ Screen annually for hypercalcaemia and serum creatinine for renal function.</li> <li>➤ Consider coeliac testings and TFTS if symptomatic.</li> <li>➤ Advise to wear sunscreen and avoid sunshine to reduce risk of hypercalaemia</li> </ul>
EXAMINATION audio-visual	<ul style="list-style-type: none"> <li>➤ Full assessment by optician/optometrist every 2 years.</li> <li>➤ People with Williams syndrome may have hearing hypersensitivity. Assessment with referral for audiology masking if concerns about hyperacusis.</li> </ul>
abdominal	<ul style="list-style-type: none"> <li>➤ Examine the abdomen for constipation.</li> <li>➤ Screen for coeliac disease and diverticular disease if symptomatic.</li> <li>➤ Renal tract ultrasound every 5 years for nephrocalcinosis</li> </ul>
Cardio-vascular	<p>Congenital heart defects (especially supra-ventricular aortic stenosis (SVAS) and peripheral pulmonary artery stenosis).</p> <ul style="list-style-type: none"> <li>➤ Full cardiovascular assessment including scans and BP (blood pressure) measurement in both upper limbs.</li> <li>➤ Echocardiogram every 5 years throughout life.</li> </ul>
MSK	<ul style="list-style-type: none"> <li>➤ Weigh annually, and avoid excessive weight gain—encourage an 'active' lifestyle.</li> <li>➤ Assessment spine for scoliosis</li> </ul>