LIST OF CLINICAL SYNDROMES LINKED TO LEARNING DISABILITIES AND THE HEALTH ISSUES THEY PRESENT

For the purpose of assisting general practitioners and practice nurses in identifying related health care needs of individuals who have learning disabilities and specific syndromes.

For use in screening and monitoring.

Could be used in other primary and secondary health care settings.

Compiled from a variety of referenced sources by Cath Scott, Community Nurse for People with Learning Disabilities

WEST SUSSEX HEALTH AND SOCIAL CARE NHS TRUST
NORTHERN LOCALITY
JANUARY 2005
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Learning Disability: What It Means

(Adapted from Selby and York PCT Specialised Learning Disability Service)

“A state of arrested or incomplete development of mind that includes significant impairment of intelligence and social function”.

World Health Organisation

Department of Health (1991) changed the terminology from Mental Handicap to Learning Disability. Different user groups and organisations use different names, which include:

- Learning Difficulty
- Intellectual Disability
- Intellectual Impairment
- Cognitive Impairment

The term learning disability will be used for the purposes of this document.

Learning Disability includes the presence of:

- A significantly reduced ability to understand new or complex information, to learn new skills (impaired intelligence), with;
- A reduced ability to cope independently (Impaired social functioning);
- Which started before adulthood, with lasting effect on development.

Department of Health: Valuing people (2001)
At present the Community Team for People with a Learning Disability (West Sussex – North) use these factors, but they are in addition to and may be associated with an IQ of below 70

**EVIDENCE THAT MAY INDICATE A LEARNING DISABILITY**

- History of being excluded or suspended from mainstream school
- Case note entries and letters that refer to Special School history
- Previous formal assessment of level of functioning (e.g. Psychology report)
- CPA documents that evidence a learning disability
- Clinical notes of various disciplines that present a picture of difficulties in a number of areas of adaptive behaviour that would not be explained by an alternative label (i.e. not purely physical or sensory difficulties, linked to mental health issues, etc)
- Reading and math skills up to grades 3 to 6 level, rarely beyond this level
- Unable to acquire complex practical and vocational skills
- Other family members diagnosed as having a learning disability

**EVIDENCE THAT MAY NOT INDICATE A LEARNING DISABILITY**

- School exam results higher than expected, attended mainstream education without Educational Support
- Employment history showing person coping without expected levels of support
- Suggestion of onset of problems in later life (post 18 yrs)
• Variable amount of support needed – which may suggest social or mental health issues and not a learning disability

• Person themselves, carers or colleagues querying label or referral to a service for people with learning disabilities

• Holds a driving license and drives a car

• Completes complex purchases without help e.g.: house purchase

• Age appropriate development until head injury or accident

• Age appropriate development until Chronic Mental Health develops

Christopher Ball
Community Nurse Manager
IQ under 20 (in Adults, mental age below 3 years). Results in severe limitation in self

The most recent World Health Organisation publication on the “Classification of Mental and Behaviour Disorders” (ICD 10) uses the term ‘Mental Retardation’ rather than ‘Mental Handicap’ or ‘Learning Disability.’ It is defined as “a condition of arrested or incomplete development of the mind, which is especially characterised by impairment of skills manifested during the developmental period; skills which contribute to the overall level of intelligence ~ i.e. cognitive, language, motor and social abilities. Retardation can occur with or without any other mental or physical condition.”

Degrees of mental retardation are conventionally estimated by standardised intelligence tests which can be supplemented by scales assessing social adaptation in a given environment. These measures provide an approximate indication of the degree of mental retardation. The diagnosis will depend on the overall assessment of intellectual functioning by a skilled diagnostician.

Intellectual abilities and social adaptation may change over time and, however poor, may improve as a result of training and rehabilitation. Diagnosis should be based on current level of functioning.

**Mild Mental Retardation**

Approximate IQ range 50 - 69 (in Adults, mental age from 9 to under 12 years). Likely to result in some learning difficulties in school. Many adults will be able to work and maintain good social relationships and contribute to society.

**Moderate Mental Retardation**

Approximate IQ range 35 - 49 (in Adults, mental age from 6 to under 9 years). Likely to result in marked developmental delays in childhood but most can learn to develop some degree of independence in self care and acquire adequate communication and academic skills. Adults will need varying degrees of support to live and work in the community.

**Severe Mental Retardation**

Approximate IQ range 20 - 34 (in Adults, mental age from 3 to under 6 years). Likely to result in continuous need of support.

**Profound Mental Retardation**

care, continence, communication and mobility.
# Syndrome Specific List For General Practitioners

This is a guide rather than an exhaustive list

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Audiovisual</th>
<th>Endocrine</th>
<th>Psychiatric / Psychological</th>
<th>CNS</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Cerebral Palsy</strong></td>
<td>Visual Impairment</td>
<td>-</td>
<td>Depression</td>
<td>CNS</td>
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<tr>
<td></td>
<td>Hearing Impairment</td>
<td></td>
<td>Variable intellectual capacity</td>
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<tr>
<td><strong>&lt;1:500</strong></td>
<td>Orthopaedic problems</td>
<td>Genito-urinary problems</td>
<td>Incontinence</td>
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<td></td>
<td>Neuromuscular problems</td>
<td>Constipation</td>
<td>Dental problems</td>
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<td></td>
<td></td>
<td>Recurrent aspiration</td>
<td>Oesophagitis</td>
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<td></td>
<td>Gastroesophageal</td>
<td>Reflux +/- bleeding/ anemia</td>
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<td>Swallowing / eating difficulty</td>
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<thead>
<tr>
<th>Syndrome</th>
<th>Audiovisual</th>
<th>Endocrine</th>
<th>Psychiatric / Psychological</th>
<th>CNS</th>
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</thead>
<tbody>
<tr>
<td><strong>Down Syndrome</strong></td>
<td>Visual impairment (multifactorial)</td>
<td>Hypothyroidism (Annual TFT recommended)</td>
<td>Depression</td>
<td>CNS</td>
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<tr>
<td></td>
<td>cataracts</td>
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<td>Alzheimer’s type dementia</td>
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<td></td>
<td>Hearing impairment (multifactorial)</td>
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<td>(Clinical onset uncommon before 40 years)</td>
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<td></td>
<td>(Annual assessments recommended)</td>
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<tr>
<td><strong>&lt;1:700</strong></td>
<td>Congenital Heart Defects (common in 40 to 50%)</td>
<td>Atlantoaxial instability</td>
<td>Blood dyscrasias</td>
<td>CNS</td>
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<tr>
<td></td>
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<td>Skin disorders, alopecia, eczema</td>
<td>Childhood leukaemia</td>
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<td></td>
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<td>Sleep apnoea</td>
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<td>Increased susceptibility to infections</td>
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<td>Coeliac disease</td>
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<td>Most cases are sporadic; 4% due to translocation involving chromosome 21 or rarely parental mosaicism</td>
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<tr>
<td>Syndrome</td>
<td>Audiovisual</td>
<td>Endocrine</td>
<td>Psychiatric / Psychological</td>
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<tr>
<td>Prader-Willi</td>
<td>Strabismus</td>
<td>NDDM (secondary to obesity)</td>
<td>Hyperphagia, Impulse control difficulties, Self-injury</td>
<td>CNS</td>
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<tr>
<td>&lt;1:10,000-25,000</td>
<td>Myopia</td>
<td>Hypogonadism, Delayed puberty</td>
<td>Hyperphagia, Impulse control difficulties, Self-injury</td>
<td>CNS</td>
</tr>
<tr>
<td>Cardiovascular</td>
<td>Muscular/ Skeletal</td>
<td>Other</td>
<td>Atypical. Most cases are sporadic</td>
<td>Other</td>
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<tr>
<td>Fragile X</td>
<td></td>
<td>Scoliosis, Kyphosis, Hypotonia, Skin picking</td>
<td>infantile failure to thrive, then hyperphagia and severe obesity, High tolerance to pain, Decreased ability to vomit, Sleep apnoea, Osteoporosis, Undescended testes, Dental Abnormalities</td>
<td>Inheritance</td>
</tr>
<tr>
<td>&lt;1:6,000</td>
<td></td>
<td>Other</td>
<td>Atypical. Most cases are sporadic</td>
<td>Inheritance</td>
</tr>
</tbody>
</table>

- **Prader-Willi Syndrome**
  - Audiovisual: Strabismus, Myopia
  - Endocrine: NDDM (secondary to obesity), Hypogonadism, Delayed puberty
  - Psychiatric / Psychological: Hyperphagia, Impulse control difficulties, Self-injury
  - CNS: CNS

- **Cardiovascular**
  - Muscular/ Skeletal: Scoliosis, Kyphosis, Hypotonia, Skin picking
  - Other: infantile failure to thrive, then hyperphagia and severe obesity, High tolerance to pain, Decreased ability to vomit, Sleep apnoea, Osteoporosis, Undescended testes, Dental Abnormalities
  - Inheritance: Atypical. Most cases are sporadic

- **Fragile X Syndrome**
  - Audiovisual: Visual impairment, Hearing impairment, Recurrent ear infections
  - Endocrine: Connective tissue dysplasia, Scoliosis, Congenital Hip Dislocation
  - Psychiatric / Psychological: Attention deficit/hyperactivity, Variable intellectual capacity, Disabled in social functioning
  - CNS: CNS

- **Cardiovascular**
  - Muscular/ Skeletal: Aortic dilation, Mitral Valve prolapse (related to connective tissue dysplasia), Connective tissue dysplasia, Scoliosis, Congenital Hip Dislocation
  - Other: Hernia (CT related), X linked Abnormalities of speech and language
  - Inheritance: X linked
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<thead>
<tr>
<th>Syndrome</th>
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<th>Endocrine</th>
<th>Psychiatric / Psychological</th>
<th>CNS</th>
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<tr>
<td>Angelmann Syndrome</td>
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<td>Glaucoma</td>
<td>Easily excitable</td>
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<td>Hyperactive</td>
<td>Severe developmental</td>
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<td>delay Epilepsy</td>
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<td>&lt;1:10,000</td>
<td>Cardiovascular</td>
<td>Muscular/</td>
<td>Joint contractures</td>
<td>Speech impairment</td>
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<td>Skeletal</td>
<td>and scoliosis (in adults)</td>
<td>Movement and balance</td>
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<td>disorder Characteristic</td>
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<td>EEG changes</td>
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<td>Variety of genetic</td>
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<td>Williams</td>
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<td>Hyperacusis</td>
<td>Variable intellectual</td>
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<td>Cardiovascular</td>
<td>Muscular/</td>
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<td>Perceptual and motor</td>
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<td>Skeletal</td>
<td>Hypertension, CVAs</td>
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<td>Chronic hemiparesis</td>
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<td>Scoliosis</td>
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<td>Hypotonia</td>
<td>Renal abnormalities</td>
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<td>Microdeletion on</td>
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<td>chromosome 7</td>
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<td>Rett</td>
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<td>Refractory errors</td>
<td>Severe intellectual</td>
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<td>&lt;1:14,000 Female</td>
<td>Cardiovascular</td>
<td>Muscular/</td>
<td>Prolonged QT Interval</td>
<td>Epilepsy Vasomotor</td>
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<td>Skeletal</td>
<td>Osteopenia</td>
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<td>Fractures Scoliosis</td>
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<td>Hyperventilation Apnoea</td>
<td>Usually sporadic X</td>
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<td>Reflux Feeding difficulties</td>
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<td>Growth failure</td>
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<td>Syndrome</td>
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<td>Psychiatric / Psychological</td>
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<tr>
<td>Noonan</td>
<td>Strabismus, refractive errors Vision/hearing impairments</td>
<td>Mild intellectual disability</td>
<td>Epilepsy</td>
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<tr>
<td></td>
<td>Cardiovascular</td>
<td>Muscular/ Skeletal</td>
<td>Other</td>
<td>Inheritance</td>
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<tr>
<td>&lt;1:10,000</td>
<td>Pulmonary Valvular Stenosis ASD, VSD, PDA</td>
<td>Scoliosis Talipes equinovarus Pectus carinatum/excavatum</td>
<td>Abnormal clotting factors, platelet dysfunction Undescended testes, deficient spermatogenesis Lyphoedenoma Hypatosplenomegaly Cubitus valgus, hand abnormalities</td>
<td>Autosomal dominant may be sporadic</td>
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<thead>
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<th>Syndrome</th>
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<th>Psychiatric / Psychological</th>
<th>CNS</th>
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<tr>
<td>Tuberous Sclerosis</td>
<td>Retinal tumours Eye rhabdomyomates</td>
<td>Variable intellectual capacity Behavioural difficulties Sleep problems</td>
<td>Cerebral astrocytomas Epilepsy</td>
<td></td>
</tr>
<tr>
<td>&lt;1:6,000 – 17,000</td>
<td>Cardiovascular</td>
<td>Muscular/ Skeletal</td>
<td>Other</td>
<td>Inheritance</td>
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<tr>
<td></td>
<td>Rhabdomyomates Hypertension</td>
<td>Bone Rhabdomyomata</td>
<td>Kidney and lung harmartomata Polycystic kidneys Liver Rhabdomyomata Dental abnormalities Skin lesions</td>
<td>Autosomal dominant</td>
</tr>
</tbody>
</table>
# Aarskog Syndrome

**ALTERNATIVE NAMES:**

- Hypertelorism Disorder
- Aarskog-Scott Syndrome

**GENERAL HEALTH SCREENING:**

- Cardiac Screening
- Orthodontic Treatment
- Hernia assessment
- Monitoring for onset of seizures
- Monitoring and assessment of respiratory function

**PHYSICAL PHENOTYPE:**

- Rounded face - Small nose with nostrils tipped forward
- Widows peak
- Short stature
- Ptosis of eyelids with Hyper telorism (Widest eyes with droopy eyelids)
- Pulmonary stenosis
- Ventricular septal defect
- Brachydactyly with syndactyly of fingers (Abnormally short fingers or toes with webbing)
- Hypoplastic Maxilla excl Malar (Underdeveloped mid-portion of the face)
- Delayed eruption of teeth
- Overriding scrotum (shawl), Undescended testicles & inguinal hernias
- Cystic changes

**PSYCHOLOGICAL / BEHAVIOURAL PHENOTYPE:**

- Learning disability - Mild

**CONTACT:**

- Aarskog Syndrome Support Group (UK)
  101 Seaview Road
  Wallasey
  Merseyside

- [WWW.aarskoginfo.co.uk](http://WWW.aarskoginfo.co.uk)
Acrodysostosis

ALTERTATIVE NAMES:
Arkless-Graham
Acrodysphasia
Maroteaux

GENERAL HEALTH SCREENING:
Hearing assessments
Monitoring and treatment for ear infections
Monitoring and treatment of skeletal changes
Regular dental checks
Dermatological assessments

PHYSICAL PHENOTYPE:
Brachycephaly (Short head measure from back to front)
Small broad upturned nose with flat nasal bridge
Protruding Jaw
Hearing Difficulties (2/3 Hearing deficit)
Short arms & Legs with deformities of the hands & Feet (cone-shaped epiphyses)
Abnormalities of the skin, genitals & skeleton (sometimes)

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
Learning Disability--80% of those affected

CONTACT:
Society of Mucopolysaccharide Diseases
46 Woodside Road
Amersham
HP6 6AJ
Email: mps@mpssociety.co.uk
Web: www.mpssociety.co.uk
www.fabry.org.uk
Aicardi Syndrome

**ALTERNATIVE NAMES:**

**GENERAL HEALTH SCREENING:**

- High Mortality in childhood.
- Speech and language assessment of swallowing difficulties
- Assessment and monitoring of epilepsy and treatment
- Ophthalmic assessment
- Monitoring and treatment of respiratory function

**PHYSICAL PHENOTYPE:**

- Affects females only
- Partial or complete absence of Corpus Callosum
- Infantile Spasm/convulsions
- Lesions of the retina of the eye
- Microcephaly
- Proencephalic cysts (Cerebrospinal fluid-filled cavities or gaps in the brain)
- Possible Scoliosis
- Cleft lip & palate.
- Brain abnormalities
- Deformities of the hands

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**

- Severe Learning Difficulties
- Communication
- Self-Injurious Behaviour (may be involuntary)
- Disturbed sleep Pattern - Seizures & waking in the night.
- Absent expressive speech

**CONTACT:**

- Contact a Family
- Tottenham Court Road
- London
- W1P 0HA
- Tel: 020 73833555
# Alpha-Thalassaemia X linked Mental Retardation Syndrome

**ALTERNATIVE NAMES:**

X-linked alpha – thalassemia mental retardation syndrome (ATRX)

**GENERAL HEALTH SCREENING:**

Nutritional advice
Assessment and monitoring for epilepsy
Monitoring and treatment for urinary tract infections
Speech and language therapy assessment/swallowing
Regular dental checks

**PHYSICAL PHENOTYPE:**

<table>
<thead>
<tr>
<th><strong>Short stature</strong></th>
<th><strong>PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>Distinctive craniofacial features:</td>
<td>Mild to severe learning disabilities</td>
</tr>
<tr>
<td>Small head circumference</td>
<td>Absent speech</td>
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<tr>
<td>Telecanthus or ocular</td>
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<tr>
<td>Hypertelorism</td>
<td></td>
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<tr>
<td>Small nose</td>
<td></td>
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<tr>
<td>Tented upper lip</td>
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<tr>
<td>Everted lower lip</td>
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<tr>
<td>Urogenital anomalies ranging from</td>
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<tr>
<td>Hypospadias and undescended testicles to</td>
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<td>Severe hypospadias and ambiguous genitalia</td>
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<tr>
<td>- prone to urinary tract infections</td>
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<tr>
<td>Cardiovascular abnormalities in some cases</td>
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<tr>
<td>Epilepsy</td>
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<tr>
<td>Skeletal abnormalities can be quite diverse</td>
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<tr>
<td>Uncoordinated swallow</td>
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<td>Constipation and poor bladder and bowel control</td>
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<tr>
<td>Mild form of hemoglobin H disease</td>
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</tbody>
</table>

**CONTACT:**

Contact a Family
Tottenham Court Road
London
W1P 0HA
Tel: 020 73833555
Angelman’s Syndrome

ALTERNATIVE NAMES:
Happy Puppet Syndrome

GENERAL HEALTH SCREENING:
Monitoring and treatment of epilepsy
Monitor Respiratory Tract Infections & Otis Media
Monitoring and advice with regards to obesity

PHYSICAL PHENOTYPE:
Movement or Balance Disorder-usually ataxia of gait &/or tremulous movement of limbs (puppet like)
Hypermotoric Movement.
Truncal Hypotonia and Limb Hypotonia with brisk reflexes, tendency to walk stiff legged, wide based gait
Characteristic posture of the arms, upheld with flexion at the wrists and elbows. Hand flapping occurs when walking
Scoliosis 10%
Possible: Microcephaly-80%
Epilepsy.
Associated: Strabismus
Hypopigmented skin & eyes
Tongue thrusting
Suck/ Swallowing Disorders
Feeding Problems in Infancy
Frequent Drooling, Protruding Tongue
Excessive Chewing/Mouthing Behaviour
Hyperactive tendon.
Feeding Problems may including
Gastroesophageal Reflux
Large Mouth with wide spaced teeth
Long Face and prominent Jaw
Thin upper lip
Mid-face Hypoplasia
Deep eyes
Flat occiput
Obesity
Speech Impairment

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
Severe Learning Disability
Behavioural Uniqueness: Any combination of frequent Laughter / Smiling.
Apparent happy demeanour, easily excitable personality, often with hand flapping movements.
Short Attention Span.
Poor social interaction skills
Typically described as sociable and affectionate
Hyperactivity
Sleep Disorders-Need for less sleep is apparent & characterised by abnormal sleep/wake cycles.

CONTACT:
ASSERT (Angelman syndrome support education and research trust)
P.O. 505
Sittingbourne
Kent
ME1 0NE
Tel: 01980 625616
### Aniridia – Wilms tumour Association

**ALTERNATIVE NAMES:**
- Familial Aniridia (AN1)
- Miller Syndrome (Sporadic non familial aniridia (AN2)
- WAGR (Wilms Tumour Aniridia Genitourinary abnormalities/ gonadoblastoma and Retardation)
- Gillespie Syndrome (AN3)

**GENERAL HEALTH SCREENING:**
- Ophthalmic assessment
- Renal Ultrasound-Wilms Tumour Cystic Lesions
- Genitalia-Assessment
- Monitoring of renal function
- Monitoring and treatment of respiratory function

**PHYSICAL PHENOTYPE:**
- Long narrow face
- Prominent nose & low set
- Poorly lobulated ears
- Palperal fissures are short & downward slanted
- Possible Growth failure
- Microcephaly
- Delayed closure of the anterior fontanel
- Tracheomalacia (Eroding of the trachea caused by excessive pressure from a cuffed dotracheal tube
- Externally or internally pseudohermaphroditism, cryptorchihism, hypospadias, renal ureteral and uterine malformations streaked ovaries, also there may be duplications or agenesis with in the genital urinary system.
- Gonadblastoma-Tumour of the testes or ovary

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**
- Learning Disability
- Epilepsy
- Attention deficit disorder
- Obsessive compulsive disorder
- Pervasive development disorder
- Autism

**CONTACT:**
- Contact a Family
  - Tottenham Court Road
  - London
  - W1P 0HA
- Tel: 020 73833555
Apert’s Syndrome

ALTERNATIVE NAMES:
Acrocephalo-syndactyly type 1
Blue Diaper syndrome
Vogtcephalo-syndactyly

GENERAL HEALTH SCREENING:
Hearing Assessment
Monitoring and treatment of ear infections
Cardiac & Renal Screening.

PHYSICAL PHENOTYPE:
Craniosynostosis- (premature fusion of the cranial sutures)
Misshapen head
High forehead
Flat occiput
Widely spaced eyes with downward slant
Flat midface & nasal bridge
Severe syndactyly
Limb abnormalities
Possible Cleft palate.
Speech Difficulties
Associated: Hydrocephalus
Hearing loss
Tooth Abnormalities
Heart & Kidney anomalies
(occasional)
Cardiac and gastrointestinal malformations

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
Some indication for memory and attentional deficits
Possible reduced intellectual capacity in some

CONTACT:
Craniofacial Support Group
www.craniofacio.org.uk/
**Bannayan- Riley-Ruvalcaba Syndrome**

**ALTERNATIVE NAMES:**
Bannayan-Zonana syndrome  
Ruvalcaba-Myhre-Smith Syndrome  
Riley-Smith Syndrome  
(Condition is the combination of 3 formerly recognised separate disorders)

**GENERAL HEALTH SCREENING:**
Ophthalmic assessment and treatment  
Dermatological assessment

**PHYSICAL PHENOTYPE:**
- ↑ Birth weight & length  
- Macrocephaly—Abnormally large head  
- Hamartomas occurring below skin surface  
- Pigmentation may appear marbled or freckled-like on the penis or vulva  
- Eye abnormalities  
- Ocular hypertelorism—Widely spaced eyes  
- Exotropia—Deviation of one eye away from the other  
- Pseudopapilledema—Abnormal elevation of the optic disc (appears swollen)  
- Hypotonia—Diminished muscle tone  
- Hamartomatous ploys may develop within the intestines.

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**
Normal to mild learning disabilities.
# Batten's Disease

<table>
<thead>
<tr>
<th><strong>ALTERNATIVE NAMES:</strong></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Neuronal ceroid lipofuscinosis</td>
<td></td>
</tr>
<tr>
<td>Juvenile Batten Disease</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th><strong>GENERAL HEALTH SCREENING:</strong></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Ophthalmic assessment</td>
<td></td>
</tr>
<tr>
<td>Monitoring epilepsy and treatment</td>
<td></td>
</tr>
<tr>
<td>Monitoring of mental health</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th><strong>PHYSICAL PHENOTYPE:</strong></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Ataxia (Gradual onset)</td>
<td></td>
</tr>
<tr>
<td>Seizures (myoclonic/tonic)</td>
<td></td>
</tr>
<tr>
<td>Retinal degeneration</td>
<td></td>
</tr>
<tr>
<td>Associated Complications:</td>
<td></td>
</tr>
<tr>
<td>Spasticity</td>
<td></td>
</tr>
<tr>
<td>Visual impairment</td>
<td></td>
</tr>
<tr>
<td>Kyphosioliosis</td>
<td></td>
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</tbody>
</table>

<table>
<thead>
<tr>
<th><strong>PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:</strong></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Learning Disabilities (Gradual Intellectual Decline)</td>
<td></td>
</tr>
<tr>
<td>Associated Complication:</td>
<td></td>
</tr>
<tr>
<td>Psychosis</td>
<td></td>
</tr>
</tbody>
</table>

**CONTACT:**

- Batten Disease Family Association
  [www.bdfauk.freeserve.co.uk](http://www.bdfauk.freeserve.co.uk)

- [www.RNIB.org.uk](http://www.RNIB.org.uk) – Visual impairments

- Batten support and Research Trust
  [www.bsrt.org.uk/index.htm](http://www.bsrt.org.uk/index.htm)
# Borjeson-forssmann-lehmann syndrome

## ALTERNATIVE NAMES:
- BORJ
- BFLS
- Borjeson syndrome
- Mental deficiency – epilepsy- endocrine syndrome

## GENERAL HEALTH SCREENING:
- Nutritional advice in relation to obesity
- Ophthalmic support
- Monitoring and treatment of epilepsy
- Cardiac assessment

## PHYSICAL PHENOTYPE:
- Obesity
- Gynecomastia
- Long thick ears
- Protruding tongue
- Hypotonia
- Hypogonadism-small penis/undescended testes
- Cataracts or other eye anomalies
- Tapering fingers
- Microcephaly
- Epilepsy
- Subcutaneous swelling gives a round fat facial appearance
- Blepharoptosis
- Dilated ventricles of the heart

## PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
- Learning disability

## CONTACT:
Genetic Alliance  
4301 Connecticut Avenue  
NW, Suite 404  
Washington DC  
20008-234  
USA  
Tel: 2029665557  
[www.geneticalliance.org](http://www.geneticalliance.org)
# Branchio –Oculo-Facial Syndrome

## ALTERNATIVE NAMES:
- BOFS
- Branchioocularfacial Syndrome
- Hemangiomatous Brachial Cleft-Lip Pseudocleft Syndrome

## GENERAL HEALTH SCREENING:
- Ophthalmic assessment
- Hearing assessment
- Monitoring and assessment of renal function
- Dermatological assessment

## PHYSICAL PHENOTYPE:
- Absent or hypoplastic thumbs/pollicies
- Anophthalmia
- Auricular pits/fistulas, auricular tags
- Blocked/absent nasolacrimal duct
- Branchial cleft/sinus/cysts
- Capillary haemangioma, cavernous
- haemangioma, thymus, general abnormalities.
- Cerebellar abnormalities (structural) Cleft palate, Cleft upper lip (non-midline)Clomboma involving optic nerve. Colomboma of retina/choroid, cataract. Dysplastic ears, deafness, coductive. Dystopia canthorum (telecanthus).Flat nose, uplift of ear lobule
- Microcephaly, microphthalmia
- Palpevral fissures slant up
- Patchy aplasia/hypoplasis of skin peri-orbital tumours/cysts
- Pits of lower lip
- Premature greying of hair
- Ptosis of eyelids
- Renal agensis
- Scalp tumours
- Short palpebral fissures, short philtrum
- Stature/length short proportionate
- Skin atrophy - patchy
- Small/yhypoplastic/deepset nails/claws
- Telangiectasia/angiokeratomata of skin

## PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
- Mild learning disability in some cases

## CONTACT:
[WWW.ABILITY.ORG.UK](http://WWW.ABILITY.ORG.UK)
# Cornelia de Lange Syndrome

## Alternative Names:
- De- Lange Syndrome
- Cornelia de Lange Syndrome
- Amsterdam Dwarfism

## General Health Screening:

<table>
<thead>
<tr>
<th>Screening Type</th>
<th>Details</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hearing Assessment</td>
<td>Monitoring of Male Genitalia</td>
</tr>
<tr>
<td>Cardiovascular Assessment</td>
<td>Hearing assessment</td>
</tr>
<tr>
<td>Ophthalmic Assessment</td>
<td>Gastroesophogeal assessment</td>
</tr>
<tr>
<td>Monitoring of Respiratory function</td>
<td></td>
</tr>
<tr>
<td>Monitoring and treatment of respiratory infections</td>
<td></td>
</tr>
</tbody>
</table>

## Physical Phenotype:

- Short Stature
- Hypertrichosis (Hirsutism)
- Synophrys
- Long curly Lashes
- Anteverted Nostrils/ Small upturned nose
- Depressed Nasal Bridge/Long Philtrum
- Thin Upper Lip
- Microcephaly
- Low set ears / hearing abnormalities
- Limb Anomalies-Partial
- Syndactyl&Miniturisation
- Associated: Eye problems—Myopia, Ptosis, or nystagmus.
- High arched palate
- Micrognathia
- Widely spaced teeth
- Facial features may change with age especially in affected males
- Less Frequent Occurrence: Congenital Heart Defects
  - Hiatus Hernia
  - Pyloric Stenosis—Stomach
  - Brachoesophagus—Abnormally Short
  - Undescended Testes
- Associated: Eye problems—Myopia, Ptosis, or nystagmus.
- High arched palate
- Micrognathia
- Widely spaced teeth
- Facial features may change with age especially in affected males
- Less Frequent Occurrence: Congenital Heart Defects
  - Hiatus Hernia
  - Pyloric Stenosis—Stomach
  - Brachoesophagus—Abnormally Short
  - Undescended Testes

## Psychological/Behavioural Phenotype:

- Moderate – Severe Learning Disabilities
- Delayed or Limited Speech development (or Absence)
- Autistic Features-Ability to relate socially
  - Physical contact Avoidance
- Little reaction to sound or pain excessive sensitivity to sensory input
- Repetitive & stereotypical movements such as Twirling
- Rigidity & Inflexibility to change
- Self-injurious Behaviour & Aggression have been noted possibly due to discomfort or frustration most often related to gastrointestinal reflux, which occurs frequently in this Syndrome.

## Striking characteristics of the syndrome, Behaviours tend to be Stereotyped & performed repeatedly.
- Temperament is significantly improved after treatment for reflux vomiting.

## Contact:
- Cornelia de lange foundation uk, Tall Trees, 106 Lodge Lane, Grays, Essex, RM16 2UL, email: info@cdls.org.uk
- Web: www.cdls.org.uk
# Charge syndrome

### ALTERNATIVE NAMES:

**CHARGE** - the name is derived from the first initial of the most commonly featured conditions with the syndrome

### GENERAL HEALTH SCREENING:

- Ophthalmic investigations - including light sensitive observations
- Hearing assessment
- Monitoring and treatment of ear infections
- Cardiology assessment and surgery if necessary
- Monitoring respiratory function
- Monitoring Genito-urinary function
- Monitoring and treatment of urinary tract infections

### PHYSICAL PHENOTYPE:

- **Coloboma** - significant vision loss especially visual fields, acuity. Sensitivity to light
- 40% facial palsy may be accompanied by sensorineural hearing loss
- 30% swallowing problems
- **Heart** defects - varying degree and severity
- **Atresia** of the choanae - often surgically repaired after birth. Respiratory problems
- Retarded growth 80% of babies
- **Genital**/urinary abnormalities - small penis and or undescended testes. Small labia. Kidney urinary tract abnormalities especially reflux
- **Ear** abnormalities - short wide ears with little or no ear lobe. Often with a snipped off appearance to the helix. Soft ears due to floppy cartilage. 80-85% have varying degree of hearing loss. Prone to ear infections and balance can be disturbed

### PSYCHOLOGICAL /BEHAVIOURAL PHENOTYPE:

- Learning disability to varying degrees

### CONTACT:

- CHARGE family support group
- 82 Gwendoline Avenue
- Upton Park
- London
- E13 0RD
- Tel: 020 8552 6961
- Email: levey2000@aol.com
- Web: [www.widerworld.co.uk/charge](http://www.widerworld.co.uk/charge)
Chromosome 3q duplication syndrome

ALTERNATIVE NAMES:
3q syndrome
3q duplication
chromosome 3q trisomy syndrome
dup (3q) syndrome
duplication 3q syndrome
partial trisomy 3q
trisomy 3q

GENERAL HEALTH SCREENING:
Cardiac assessment and monitoring
Monitoring and treatment of epilepsy

PHYSICAL PHENOTYPE:
Microcephaly
Brachycephaly
Micrognathia
Square face and full cheeks
Low set ears and malformed ears
Hypertelorism
Epicanthal folds prominent philtrum
Broad nasal root
High arched palate-possible cleft lip or palate
Flattened chest with widely spaced nipples
Omphalocele
Clindactyly
Brachydactyly
Camptodactyly and tapered fingers
Dermatoglyphic defects
Hypotonia
Hypertrichosis and hypoplastic nails
Epilepsy
Craniostenosis
Heart abnormalities – septal defects

PSYCHOLOGICAL/BEHAVIORAL PHENOTYPE:
Learning disability

CONTACT:
Contact a Family
Tottenham Court Road
London
W1P 0HA
Tel: 020 73833555
Chromosome 4q Duplication Syndrome

ALTERNATIVE NAMES:
4q+ Syndrome
4q duplication Syndrome
chromosome 4q Trisomy
Dup(4q) Syndrome
Duplication 4q syndrome
Partial trisomy 4q
Trisomy 4q

GENERAL HEALTH SCREENING:
Ophthalmic assessment
Monitoring and assessment of cardiac function
Monitoring and assessment of renal function
Hearing assessment
Monitoring and treatment of epilepsy

PHYSICAL PHENOTYPE:
Microcephaly
Microganthia
Prominent metopic suture
Malformed, low set ears with prominent anthelices
Hypertelorism
Strabismus
Narrow downslanting, palpebral fissures
Shallow nasal bridge, straight nasofrontal angle, short philtrum and epicanthal folds
Umbilical or inguinal hernia
Rockerbottom feet, thumb deformities, syndactyly, abnormal dermatoglyphics
Scoliosis
Hypertonia or hypotonia
Epilepsy
Heart murmur
Venous return anomalies
Horseshoe kidney
Renal hypoplasia
Urethro-vesical reflux
Cripotorchidism
Inguinal hernia

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
Learning Disability

CONTACT:
Contact a Family
Tottenham Court Road
London
W1P 0HA
Tel: 020 73833555
Chromosome 9q Duplication Syndrome

**ALTERNATIVE NAMES:**

9q+ Syndrome
9q Duplication Syndrome
Chromosome 9q Trisomy
Dup (9q) Syndrome
Duplication 9q Syndrome
Partial Trisomy 9q
Trisomy 9q

**GENERAL HEALTH SCREENING:**

Monitoring and assessment of cardiac function
Dermatological assessment
Ophthalmic assessment
Hearing assessment

**PHYSICAL PHENOTYPE:**

Microcephaly, dolichocephaly, retroganthia, bulging forehead and facial asymmetry
Large pinnae and crumpled helices
Narrow palpebral fissures, blepharoptosis
Deeply set eyes microthalmia, strabismus and occasional ectopia lentis
Torticollis
Joint contractures
Scoliosis and kyphosis
Erythema
Encephalopathy and brisk reflexes
Mitral and aortic regurgitation and systolic click
Cryptorchidism and hypoplastic scrotum

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**

Learning Disability
Speech difficulties

**CONTACT:**

UNIQUE (Rare chromosome disorder support group)
P.O. Box 2189
Caterham
CR3 5GN

Tel: 01883 330766
Email: info@rarechromo.org
Web: www.rarechromo.org
Coffin-Lowry Syndrome

ALTERNATIVE NAMES:
Coffin Syndrome

GENERAL HEALTH SCREENING:
Cardiac screening
Hearing assessment

PHYSICAL PHENOTYPE:
Craniofacial abnormalities: Maxillary hypoplasia
Abnormally prominent brow
Palperbral fissures
Hypertelorism, large ears and/or unusually thick eyebrows
Large soft hands with short tapered fingers
Short stature
Kyphoscoliosis
Unusual prominence of the sternum
Delayed bone development
Hypothurnar crease
Midline lingual furrow
Malocclusion
Hypodontia
Narrow intervertebral spaces
Cardiomyopathy or mitral valve stenosis
Sensory defensiveness (most noticed in early childhood)

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
Learning disability

CONTACT:
Coffin-lowry Syndrome foundation
www.clsf.info/
# Coffin Siris Syndrome

**ALTERNATIVE NAMES:**
- Dwafism - onychodysplasia syndrome
- Fifth digit syndrome

**GENERAL HEALTH SCREENING:**
- Assessment and monitoring of cardiac function
- Monitoring and treatment of respiratory infections

**PHYSICAL PHENOTYPE:**
- Short stature
- Abnormal 5th digit (malformed, missing, or underdeveloped terminal phalanx-visible on x-ray)
- The finger nail is also usually underdeveloped or missing - other digits may also be similarly affected
- Flat/low/wide nasal bridge
- Anteverted and wide nasal tip
- Wide or large mouth with thick prominent lips
- Long eye lashes (hypertichosis)
- Hairy body and face (hirsutism)
- Thick eye brows
- Sometimes sparse scalp hair
- Associated features:
  - Widely spaced eyes
  - Ptosis
  - Microcephaly
  - Low set ears
  - Short philtrum
  - Prominent or wide forehead
  - Balbous nose
  - Occassionally congenital heart defect
  - Frequent respiratory infections during infancy
  - Stomach ulcers
  - Eczema

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**
- Mild – severe learning disability
- Situation specific maladaptive behaviours

**CONTACT:**
- Contact a Family
- Tottenham Court Road
- London
- W1P 0HA
- Tel: 020 73833555
# Cohen Syndrome

## Alternative Names:

- Cohen Syndrome

## General Health Screening:

- Dietetic assessment
- Ophthalmic assessment
- Assessment and monitoring of epilepsy
- Maxillofacial assessment

## Physical Phenotype:

- Microcephaly
- Short philtrum
- Prominent upper central incisors
- Palpebral fissures slant down
- Narrow and high arched palate
- Microganthia
- Maxillary hypoplasia
- Truncal obesity
- Short stature
- Arachnodactyly
- Narrow hands and feet
- Joint laxity muscle weakness/myopathy
- Ophthalmic irregularities, myopia strabismus
- Pigmentary retinopathy/chorioretinitis
- Epilepsy

## Psychological/Behavioural Phenotype:

- Learning Disability

## Contact:

Cohen Syndrome support group
45 Compton Way
Middleton Junction
Middleton
Manchester
M24 2BU

Tel: 0161 653 0867
Email: cohensyndrome@hotmail.com
Cockayne Syndrome

ALTERNATIVE NAMES:
Deafness-Dwarfism-retinal atrophy
Dwarfism with renal atrophy and deafness
Neill-Dingwall Syndrome
Progeroid Nanism

GENERAL HEALTH SCREENING:
Physiotherapy
Dermatological assessment
Ophthalmic assessment
Hearing assessment
Ultrasound examination for renal abnormalities

PHYSICAL PHENOTYPE:
Short stature decreased subcutaneous fat
Progressive kyphosis
Relatively long limbs
Sensitivity to sun light
Appearance of premature aging
Cerebral dysfunction with ataxia and or tremor
Hydrocephalus with progressive ataxia and urinary incontinence
Microcephaly
Spasticity with reduced tendon reflexes
Visual: corneal opacities
Impaired lacrimation
Cataracts
Speckled pigmentation of the fundus
Retinitis pigmentosa-retinal atrophy
Optic atrophy-decreased visual acuity,blindness
Sensitivity to sunlight
Facial rash
Scaly skin, dyspigmentation, scarring atrophy of exposed areas
Hearing impairment
Dental caries
Renal disease

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
Learning disability
## Cri-Du-Chat Syndrome

### ALTERNATIVE NAMES:
- Deletion 5p Syndrome
- Chromosome 5 short arm deletion

### GENERAL HEALTH SCREENING:
- ENT assessment
- Ophthalmic assessment
- Assessment and monitoring of cardiac function
- Psychology assessment
- Hearing assessment

### PHYSICAL PHENOTYPE:
- Microcephaly
- Broad based nose
- Microganthia
- Low set poorly formed ears
- Malocclusion
- Abnormal palate
- Round face
- Preauricular tag - possible
- Hypertelorism
- Epicanthal folds down slanting or oblique palpebral fissures
- Strabismus
- Short stature
- Simian crease
- Distal axial triradius
- Short metacarpal or metatarsals
- Pes planus
- Partial syndactyly
- Infantile hypotonia
- Abnormal Larynx
- Cat like cry - often lost by the age of two
- Feeding difficulties in infancy
- Diastasis recti
- Inguinal hernia
- Short neck
- Congenital heart disease
- Striking facial appearance - features show a progression with age apparent coarseness in adolescence

### PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
- Learning Disability
- Communication difficulties
- Hyperactivity, restlessness, irritability, and a low threshold for frustration, destructiveness and self stimulator behaviour: hand sucking and head banging

### CONTACT:
- Cri-du-chat syndrome support group
  - Penny Lane
  - Barwell
  - Leicestershire
  - LE9 8HJ
  - Tel: 01455 841680
  - Web: [www.criduchat.u-net.com](http://www.criduchat.u-net.com)
- 5p minus society – family support group
  - Web: [www.fivepminus.org](http://www.fivepminus.org)
# Crouzon's Syndrome

## Alternative Names:
- Craniosynostosis Syndrome

## General Health Screening:
- Assessment and management of epilepsy
- Assessment and treatment of headaches
- Assessment and management of hearing
- Ophthalmic assessment
- Respiratory assessment

## Physical Phenotype:
- Cranio-facial abnormalities including:
  - Acrocephaly (cone shaped head), brachycephaly, palable ridging, flat occiput,
  - High prominent forehead, flattened face with maxillary hypoplasia, relative mandibular
  - Prognathism, low set ears - conductive hearing loss, downslanding palperbral fissure,
  - Exophthalmos with shallow orbits, iris coloboma, ptosis.
- Exposure to conjunctivitis or keratitis
- Decreased visual acuity, class III malocclusion with maxillary crowding
- Hypertelorism
- Strabismus
- Nystagmus
- Deviated nasal septrum
- Short upper lip
- High arched narrow palate
- Spine abnormalities
- Subluxion of the radial heads

## Psychological/Behavioural Phenotype:
- Frequent headaches
- Mild to moderate learning disability
- Epilepsy

## Contact:
- Headlines: The Craniofacial Support Group
  128 Beesmoor Road
  Frampton Cotterell
  Bristol
  NS36 2JP
- Tel: 01454 850557
- Email: info@headlines.org.uk
- Web: www.headlines.org.uk
Deletion 3p syndrome

**ALTERNATIVE NAMES:**

3p syndrome  
3p deletion syndrome  
chromosome 3p monosomy  
del (3p) syndrome  
deletion 3p syndrome  
partial monosomy 3p

**GENERAL HEALTH SCREENING:**

Auditory assessment  
Ophthalmic assessment  
Physiotherapy assessment  
Monitoring and assessment of cardiac function  
Genito-urinary assessment

**PHYSICAL PHENOTYPE:**

Microcephaly  
Dolichocephaly  
Micrognathia  
Low set malformed ears with dysmorphic pinnae and preauricular pits  
Blepharoptosis  
Iris coloboma  
Upslanting palpebral fissures  
Hypertelorism  
Synphrys  
Prominent nasal bridge  
Long philtrum  
Epicanthal folds  
Downturned mouth thin lips  
Umbilical hernia  
Ulnar deviation of hands  
Clinodactyly/polydactyly  
Rocker bottom feet  
Abnormal insertion of thumbs  
Hypotonia  
Heart murmur  
Cryorchidism  
Hypoplastic bladder/urethers  
Hypogonadism  
Kidney cysts

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**

Learning disability

**CONTACT:**

UNIQUE – Rare Chromosome Disorder Support Group  
P.O. Box 2189  
Surrey  
CR3 5GN  
Tel: 01883 330766  
Web: [www.rarechromo.org](http://www.rarechromo.org)
Deletion 4q syndrome
(about 70 percent die within the first two years of life)

**ALTERNATIVE NAMES:**

- 4q syndrome
- 4q deletion syndrome
- chromosome 4q syndrome
- del (4q) syndrome
- monosomy 4q
- partial monosomy 4q

**GENERAL HEALTH SCREENING:**

- Monitoring and treatment of epilepsy
- Auditory assessment
- Monitoring of possible pneumonia
- Renal assessment
- Cardiac assessment and monitoring

**PHYSICAL PHENOTYPE:**

<table>
<thead>
<tr>
<th>Cardiovascular defects</th>
<th>Learning disability</th>
</tr>
</thead>
<tbody>
<tr>
<td>Possible cleft lip/palate</td>
<td></td>
</tr>
<tr>
<td>Micrognathia</td>
<td></td>
</tr>
<tr>
<td>Macrocephaly</td>
<td></td>
</tr>
<tr>
<td>Facial asymmetry</td>
<td></td>
</tr>
<tr>
<td>Low set posteriorly angulated ears with deformed helices and stenotic ear canals</td>
<td></td>
</tr>
<tr>
<td>Hypertelorism</td>
<td></td>
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<tr>
<td>Short nose with depressed bridge</td>
<td></td>
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<tr>
<td>Absent digits clinodactly with tapered 5th fingers and over lapping toes</td>
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<tr>
<td>Restricted elbow movement and hip dislocation</td>
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<tr>
<td>Vertebral anomalies</td>
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<tr>
<td>Hypotonia</td>
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<tr>
<td>Hydrocephalus</td>
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<tr>
<td>Pneumonia</td>
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<tr>
<td>Anterior anus and jejunal atresia</td>
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<tr>
<td>Cystic and rotated kidneys</td>
<td></td>
</tr>
<tr>
<td>Epilepsy</td>
<td></td>
</tr>
</tbody>
</table>

**CONTACT:**

Contact a Family
Tottenham Court Road
London
W1P 0HA
Tel: 020 73833555
# Deletion 18q syndrome

## ALTERNATIVE NAMES:
- Degrouchy syndrome
- Carp mouth syndrome

## GENERAL HEALTH SCREENING:
- Ophthalmic assessment
- Assessment and monitoring of epilepsy
- Oncology assessment
- Auditory assessment

## PHYSICAL PHENOTYPE:
- Microcephaly
- Short stature
- Protruding chin
- Fish like mouth
- Cleft lip/palate
- Ocular: strabismus, nystagmus
- Genital hypoplasia
- Congenital heart defects
- Fingers are long and spindle shaped
- Dermatological: high frequency of whorls
- Mild obesity ataxia hypotonia
- Seizures
- Malformations of pulmonary veins
- Thyroid carcinoma
- Cerebella astrocytoma
- Cortical nephroblastomatosis

## PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
- Learning disability
- Hyperactive and aggressive behaviour

## CONTACT:
- UNIQUE – Rare Chromosome Disorder Support Group
  - P.O. Box 2189
  - Surrey
  - CR3 5GN
  - Tel: 01883 330766
  - Web: [www.rarechromo.org](http://www.rarechromo.org)
Deletion 11q deletion syndrome

ALTERNATIVE NAMES:

Jacobsen syndrome
11q-syndrome
chromosome 11q monosomy
del (11q) syndrome
monosomy
partial monosomy 11q

GENERAL HEALTH SCREENING:

Cardiac assessment and monitoring
Genito-urinary assessment
Ophthalmic assessment

PHYSICAL PHENOTYPE:

Microcephaly
Retroganthia
Low set dysmorphic ears
Strabismus
Colboma
Blepharoptosis
High arched palate
Diastasis of therectus abdominis muscle
Camptodactly
Syndactly
Single palmar crease
Hypertonia or hypotonia
Hepatomegaly
Cardiac defects
Cryptorchidism
Anemia
Hearing impairment
Trigoncephaly
Epicanthus
Telecanthus
Isoimmune thrombocytopenia

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Mild-severe learning disability
Speech and language difficulties

CONTACT:

Contact a Family
Tottenham Court Road
London
W1P 0HA

Tel: 020 73833555
Down's Syndrome

ALTERNATIVE NAMES:
Chromosome 21 Syndrome
21 Trisomy
Langdon Down Syndrome
Morbus Down
 Congenital acromicria Syndrome
Trisomy 21
Trisomy 21 Syndrome

GENERAL HEALTH SCREENING:
Monitoring and assessment of cardiovascular function
Hearing assessment
Monitoring and assessment of thyroid function

PHYSICAL PHENOTYPE:
Flattened broad facies, flat occiput,
brachycephaly and small maxilla
Abnormal auricles and absent lobules
Upslanting eyes
Palpebral fissures/brushfield spots
Strabismus nystagmus
Hypertelorism
Cataract
Small nose and epicanthal folds
Large protruding and fissured tongue
Open mouth, narrow plate
Thick, small and malalagned teeth
Thick and broad neck with excess skin
Petus excavatum and petus carinatum
Umbilical hernia and diastasis recti
Short, broad hands, brachymesophalangy of
the fifth fingers, clinodactyly and gap between
hallux and second toe
Hyperextensible joints and hip dysplasia
Dorsolumbar kyphosis
Hypotonia
Cardiovascular defects
Grypto orchidism, hypogonadism
Polycythemia
Hypothyroidism
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OBESITY

- Short stature is a recognised feature of most people with Down's syndrome. Appropriate growth monitoring is essential. Those who are excessively short or underweight may have additional pathology which requires investigation and treatment.

- Down's specific growth charts provide essential reference values. The possibility of additional pathology should be considered for those falling in the lower centiles who do not have congenital heart disease.

- Weight for height should be assessed using standard BMI charts.

- Overweight/obesity is not inevitable and should always be thoroughly assessed.

- Appropriate anticipatory guidance regarding diet and physical activity should be given for all those with the syndrome.

Based on DSMIG (Down’s Syndrome Medical Interest Group) guidelines for basic essential medical surveillance 31.01.01
EYES - OPHTHALMIC PROBLEMS

- There is a high incidence of ocular disorder among people with Down's syndrome (3).
- Refractive errors and/or squint may be present from an early age.
- Cataract and/or glaucoma may occur in infancy (7).
- Nystagmus is present in at least 10% of the population.
- Keratoconus is more common in adolescents and young adults. Untreated most of these disorders are a significant cause of preventable secondary handicap (8,9)
- Some deviation from normal and should be kept under close review.
- Children and adults with Down's syndrome should be expected to respond to standard testing procedures at appropriate developmental age but a distraction free environment may be necessary to optimise performance. Distance and near vision should be checked at every review.
- High Street opticians give an excellent service but subjects who are not cooperative in this should be referred to a specialist clinic.

References:

DSMIG (UK) 2001 www.dsmig.org.uk 15.01.02
HEARING IMPAIRMENT

- Well over 50% of people with Down’s syndrome have significant hearing impairment which may be mild, moderate, severe or profound.
- Sensorineural and/or conductive loss may be present at any age.
- Hearing impairment can be successfully managed in this population. 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). The natural history of OME and response to intervention differ from that in the general population hence local surveillance and management protocols need to be set up specific to people with Down’s syndrome.
- Because of an increased incidence of congenital sensorineural loss newborns should be included in neonatal screening programmes.
- At all ages people with Down's syndrome have narrow ear canals which predispose to accumulation of wax. This may affect impedance testing and hearing.
- Because of increased incidence of sensorineural as well as conductive loss the frequency range tested should include 8000Hz whenever feasible as this may be an early warning of impending sensorineural deafness.
- Diagnostic Auditory Brain Stem (ABR) responses in people with Down's syndrome must be interpreted with caution.
- In adults with the syndrome hearing assessment is essential in the differential diagnosis of depression and dementia.

References

DSMIG (UK) 2001 www.dsmig.org.uk 15.01.02
Approved by BACDA and BAAP. Sept 2000
CARDIAC DISEASE - CONGENITAL AND ACQUIRED

- Between 40 and 50% of babies with Down's syndrome have congenital heart defects. Of these 30-40% have complete atroventricular septal defects. Most AVSD can be successfully treated if the diagnosis is made and the baby referred for full corrective surgery before irreversible pulmonary vascular disease is established.

- There must be a high level of clinical suspicion of congenital heart disease for all newborns with the syndrome.

- Clinical examination alone is insufficient to detect all of even the most serious abnormalities.

- Even if early investigations are reported as 'normal', if a child develops signs or symptoms of cardiac disease appropriate investigations must take place as structural problems may not have been evident at an earlier age. Those with suspected problems should be referred for immediate cardiological review so that intervention, if necessary, can take place before pulmonary vascular disease develops.

- It is recognised that minor heart defects (atrial septal defect and small ventricular septal defects) may be missed in those children who do not have echocardiograms but these should declare themselves clinically, as for any child, in the normal course of child health surveillance.

- There is an increased incidence of mitral valve prolapse and of aortic regurgitation in adults. This has implications for infective carditis prevention particularly because of the high incidence of periodontal disease among this population. We therefore recommend an echo screen for all people with Down's syndrome early in adult life.

- If a potential risk situation for infective endocarditis arises for an adult with Down's syndrome who has not had an adult echo, preventive prophylactic measures should be started.
References:


Dr Jennifer Dennis (Chairman. DSMIG -U.K) 07.10.99
ALZHEIMER'S-TYPE DEMENTIA

- Estimates vary, but a reasonable conclusion is that 25% or more of individuals with Down syndrome over age 35 show clinical signs and symptoms of Alzheimer's-type dementia. The percentage increases with age.

- Individuals with Down syndrome are three to five times more likely than the general population to develop Alzheimer's disease.

- Symptoms of a variety of other diseases and conditions mimic the symptoms of Alzheimer's disease: personality change, decline in daily living skills, memory loss, changes in coordination and gait and other changes.

- Diseases and conditions such as depression, thyroid disorders, brain tumour, recurrent brain strokes, metabolic imbalances and various neurological conditions must be ruled out prior to a diagnosis of Alzheimer's disease.

- It is recommended that individuals with Down syndrome take a baseline test of cognitive function at age 30, and that this test be repeated annually to determine any deterioration in this function.

- Some Alzheimer's disease symptoms can be treated, although there is no current means of curing or arresting the disease.

- Current research suggests a causative link between the extra "gene dosage" from the third chromosome 21 of Down syndrome and Alzheimer's disease.

Dr. Ira Lott: Professor of Paediatrics University of California at Irvine
THYROID DYSFUNCTION

- Hypothyroidism affects 10-20% of people with Down’s syndrome. It can occur at any age. Screening blood tests are essential. It can be successfully treated (1).
- Evidence for a steady decline in thyroid function as age increases
- Congenital hypothyroidism is also over-represented (2).
- Neonatal screen followed by either: Venous blood screen - T4, TSH, and thyroid antibodies checked at age 1 and thereafter every 2 years for life. If normal T4 but mildly raised TSH or antibodies check more frequently or:
  Finger prick capillary blood screen - annual Guthrie TSH check. All with Guthrie TSH > 10mU/l to be referred for venous sampling
- Whatever the results of screening tests, clinicians to have a low threshold for testing if clinical suspicion at any time.
- Clinical pointers as in general population: - lethargy and/or changes in affect, cognition, growth and weight
- The aetiology is heterogeneous but an auto-immune type thyroiditis is often causative. Thyroid microsomal antibodies are found in around 30% of people with Down’s syndrome and have been detected in children with the syndrome as early as age 2 years. The presence of microsomal antibodies does not necessarily imply thyroid dysfunction but should be taken as an indication to check thyroxine levels frequently. The absence of antibodies does not preclude hypothyroidism from other causes.
- Hyperthyroidism, though much less frequent, is also over-represented among the population with Down’s syndrome (3).
- Differential diagnosis from depression and dementia critically important

References:

**DSMIG guidelines - Last updated 16.03.00 Down’s Syndrome Association**

**Medical Series Revised June 2000**
CERVICAL SPINE INSTABILITY

- People with Down's syndrome have a small risk for acute or chronic neurological problems caused by cervical spine instability (1.2)
- Currently there is no screening procedure which can predict those at risk. In particular cervical spine X rays in children have no predictive validity for subsequent acute dislocation/ subluxation at the atlantoaxial joint (4).
- Children with Down's syndrome should not be barred from sporting activities because there is no evidence that participation in sports increases the risk of cervical spine injury any more than for the general population (6).
- the risk of injury is small, if any child or adult needs an anaesthetic the anaesthetist and recovery room staff must always be reminded of the diagnosis so that appropriate care can be taken to avoid cervical injury whilst manipulating the head and neck in the unconscious subject (9)
- If a person with Down's syndrome develops pain behind the ear or elsewhere in the neck, abnormal head posture, torticollis, deterioration of gait, manipulative skills, or bowel and /or bladder control they should be referred immediately to an appropriate specialist (usually a neurologist or a spinal orthopaedic surgeon).

References
GROWTH

- Short stature is a recognised characteristic of most people with Down’s syndrome. Average height at most ages is around the 2nd centile for the general population. For the majority the cause of growth retardation is not known (1). Some conditions leading to poor growth - congenital

- Heart disease (3); sleep related upper airway obstruction (4); coeliac disease (5); nutritional inadequacy due to feeding problems; and thyroid hormone deficiency (7.) occur more frequently among those with the syndrome.

- Regular surveillance of growth, general health, nutritional and thyroid status should aid in early identification of pathological causes of growth retardation.

- UK/Republic of Ireland growth charts for healthy children with Down’s syndrome from birth to 18 years are available (9.10). These reference values are essential for assessing linear growth.

References:

DSMIG (UK) 2000 www.dsmig.org.uk 15.01.02
RESPIRATORY DISORDERS

- Primary cause of morbidity and/or hospital admission particularly in young children

- Little published research. Significance of problems often under recognised. Specialist investigation and treatment often necessary but often not sought.

- Increased prevalence sleep related upper airway obstruction and lower airway disease

- Underlying pathology often multifactorial

- Lower airway problems - Contributory factors include:
  - Gastro-oesophageal reflux
  - Hypotonia
  - Relative obesity
  - Immune dysfunction
  - Cardiac disease
  - Large airway compression
  - Small lower airway volume
  - Tracheobronchomalacia
  - Pulmonary hypoplasia
  - Subpleural cysts

- Upper airway problems - contributory factors include
  - Hypotonia
  - Obesity
  - mid-face hypoplasia
- relative glossoptosis
- small upper airway volume
- increased secretions
- nasal congestion
- tonsils and adenoids

- Important to recognise possibility of gastro-oesophageal reflux. Investigate as necessary and treat aggressively

- Attempt to eradicate nasal congestion
  - Mechanical decongestion - Nuk decongestor
  - Intermittent continuous low dose antibiotics
  - Trial cow’s milk free diet

- Treat chronic lower airway disease aggressively
  - Continuous prophylactic antibiotics
  - Regular inhaled corticosteroids

Mainly based on conference paper by Dr Iolo Doull at RSM conference. April 2001. DSMIG
SLEEP RELATED UPPER AIRWAY OBSTRUCTION (SrUAO).

- Occurs in up to 60% of those with Down's syndrome

- Can be caused by several different factors present in DS: the flattened midface, narrowed nasopharyngeal area, low tone of the muscles of the upper airway and enlarged adenoids and/or tonsils.

- Other disorders of breathing also found. 65-80% of children with DS have nocturnal hypoventilation and/or decreased oxygen saturation

- Most frequent clinical signs - snoring and chest wall recession. May also have abnormal sleep postures and frequent nocturnal arousals.

- May be adverse effects on daytime functioning, growth and development.

- Sleep study facilities/procedures fragmented and variable throughout UK. May need referral to specialist centre

- May exacerbate pulmonary hypertension in those with congenital heart disease.

- May lead to life threatening acute obstructive events particularly if given sedation for any reason.

- Treatment depends on the individual's clinical problem, but Ts and As may often help. Hospital admission recommended because of increased risk of post surgery airway problems

- Young children may improve with age

Based on conference paper by Dr Martin Samuels at RSM conference April 2001
COELIAC DISEASE/GLUTEN SENSITIVITY

• Prevalence 4 - 17% depending on age of sample and country of origin

• May be associated with type 1 diabetes and/or thyroid disease

• Clinical diagnosis difficult because of overlap with normal features of the syndrome therefore need to have low threshold of clinical suspicion

• AGA screen not useful in DS population. AEA effective

• AEA screen for all with major or minor symptoms as follows:
  o Disordered bowel function tending to diarrhoea or to new onset constipation
  o Failure to thrive as indicated using Down's syndrome specific reference charts (Harlow Printing 2000);
  o Abdominal distension
  o General unhappiness and misery
  o Arthritis
  o Rash suggesting dermatitis herpetiformis
  o Existing type 1 diabetes, thyroid disease or anaemia

• If AEA positive, or if negative but with significant symptoms, proceed to small intestinal biopsy as in general population.

• Treat with gluten free diet as for general population

• Dietary compliance seems no more problematic than in general population

Based on DSMIG Clinical Awareness Notes - J Dennis, C.Charlton. G Holmes
© DSMIG 8.11.01
EPILEPSY

- Recent studies report an incidence of 5-10%
- 1-3% starting either in 1st year of life or in middle age with the onset of Alzheimer's
- *Infantile spasms* are the most common type of seizures seen in infancy and usually are well controlled with either steroids or other anticonvulsants. They generally have a favourable cognitive outcome, compared with the general population.
- Tonic-clonic seizures are most commonly seen in older persons with Down syndrome, and they respond well to anticonvulsant therapy in most cases.
- The increased incidence of seizures is not thought to be solely the result of abnormal brain development, but can be related to cardiac defects, infections, and irregularities of one or more neurotransmitters.

References:

*Down Syndrome Quarterly* may be accessed through the home page of *Down Syndrome Quarterly* at "http://denison.edu/dsq."
DENTAL CARE

• Although 40 to 50% of babies with Down syndrome are born with some type of cardiac abnormality, most receive surgical correction within the first few years of life. There is, however, an abnormally large percentage who develop Mitral Valve Prolapse (MVP) by adulthood. The incidence of MVP in the normal population is between 5 - 15%. Approximately 50% of adults with Down syndrome have MVP requiring subacute bacterial endocarditis prophylaxis for dental treatment.

• A compromised immune system with a corresponding decrease in number of T cells is characteristic of most individuals with Down syndrome. This contributes to a higher rate of infections and is also a contributing factor in the extremely high incidence of periodontal disease.

• Children with Down syndrome often have chronic upper respiratory infections. These contribute to mouth breathing with its associated effects of dry mouth (xerostomia) and fissuring of the tongue and lips.

• There is also a greater incidence of aphthous ulcers, oral candida infections and acute necrotizing ulcerative gingivitis.

• Reduced muscle tone causes less efficient chewing and natural cleansing of the teeth. More food may remain on the teeth after eating due to this inefficient chewing.

• The roots of the teeth in patients with Down syndrome tend to be small and conical. This is an important factor when considering orthodontic tooth movement and also contributes to early tooth loss in periodontal disease.

References:
Dr. Pilcher is an associate professor in the College of Dental Medicine, Medical University of South Carolina, USA. This paper was originally presented at the Sixth World Congress on Down Syndrome in Madrid, Spain, in October, 1997. It was published in the journal Down Syndrome Research and Practice, Vol. 5, No. 3, p 111-116, 1998. © The Down Syndrome Educational Trust. Barnett, Friedman, & Kastner, (1988).
SEXUALLY ACTIVE WOMEN

- Cytologic screening (smear test) every 1-3 years following the age of first intercourse. For women who are not sexually active, single-finger bimanual examination with finger-directed cytology exam. Screening pelvic ultrasound every 2-3 years for women who refuse or have inadequate follow-up bimanual examinations. This may require referral a gynaecologist with experience with individuals with special needs. Otherwise, pelvic ultrasound may be considered in place of pelvic examinations.

References:
Down Syndrome Quarterly
INFECTIOUS DISEASE/IMMUNOLOGY

- A compromised immune system with a corresponding decrease in number of T cells is characteristic of most individuals with Down syndrome.

- Persons with DS who have serious recurrent respiratory and systemic infections are often evaluated for immune function.

- Intravenous gamma globulin replacement therapy should be a consideration in a person with DS who presents with serious recurrent bacterial infections and documented IgG subclass 4 deficiency. The cellular immunity deficits described in individuals with DS have the greatest documented clinical impact on gingivitis and periodontal disease.

References:

Ugazio et al. (1990). Immunological features of Down syndrome: a review. American Journal of Medical Genetics, 7(supplement), 204-212.

Down Syndrome Quarterly
BLOOD DISORDERS

There are various disorders associated with down's syndrome

- Anaemia
- Aplastic anaemia
- Polycythemia
- Neutropenia
- Leukaemia
- Myelofibrosis
- Transient myeloproliferative

References:
DEPRESSION

- The signs of depression in typical individuals usually consists of a sad, irritable mood, along with disturbances of appetite, sleep, and energy, and loss of interest in previously enjoyable activities.

- Persons with Down syndrome are more likely to present with skill and memory losses, significant activity slowdowns, and hallucinatory-like self talk and more extreme withdrawal (psychotic features).

- Persons with Down syndrome often develop depressive disorders in reaction to loss: death of a family member, change in a roommate, retirement of a caregiver from a group home, etc.

References:

Down Syndrome Quarterly
SKIN CONDITIONS

- Chelitis - presence of fissures and red, scaly skin at the corners of the mouth and lips. This is usually due to moisture collecting at the corners of the mouth.

- Atopic dermatitis

- Seborrhea

- Hyperkeratosis in people with DS occurs on the palms and soles of the feet.

- Syringomas

- Elastosis perforans serpiginosa

- Vitiligo - loss of pigmentation of the skin in well-defined areas. It may occur anywhere on the body and at any age. Vitiligo is not a common problem in people with DS, but is still more common than in the general population.

- Acanthosis Nigrans - an increase in pigmentation. The darker skin is also slightly elevated and scaly. One large study in Spain reported that out of 51 adults with DS, 26 had acanthosis nigrans. This condition most often appears on the back of the neck, the hands, and the groin. While acanthosis nigrans has been associated with type II diabetes mellitus, none of the affected adults with DS with acanthosis nigrans had evidence of diabetes.

- Folliculitis - inflammation and/or infection of hair follicles of the skin, appears as small red bumps or yellowish pustules. Most infections are due to the bacteria staphylococcus, though a fungal version has been described in adults with Down syndrome

- Scabies. For reasons unknown, this infection is a common problem in teens and adults with DS, and tends to be a worse infection than in the general population.

- Alopecia areata, believed to be due to an autoimmune process. People with DS are more prone to autoimmune diseases, such as diabetes, hypothyroidism and celiac disease. Alopecia areata is more common in people with DS, occurring in
5 to 9% of the population (compared to 1 to 2% of the general population). A gene implicated in the cause of alopecia areata has been found on the 21st chromosome.

Dermatologic Disorders in Down Syndrome by Dr. Len Leshin, MD, FAAP (2001)
Dubowitz syndrome

ALTERNATIVE NAMES:
Dwarfism-eczema-peculiar facies syndrome

GENERAL HEALTH SCREENING:
Oncology assessment                         Assessment of hearing
Ophthalmic assessment                      Dental assessment
Dermatological assessment                Gastrointestinal assessment

PHYSICAL PHENOTYPE:
Short stature
Sparse hair
Eczema
Microcephaly
Micrognathia
Prognathism triangular face
High sloping forehead
Craniosynostosis
Prominent dysplastic ears
Hypoplastic tragus and or pre-auricular fistulae
Dolichocephaly
Trionocephaly
Small narrow face
Brachycephaly
Blepharophimosis
Epicanthal folds
Variable ptosis
Chronic diarrhea/constipation
Possible rectal prolapse
Carious teeth
Leukaemia
Neuroblastoma
Aplastic anaemia
Frequent infections
Hypertelorism
Eczema/atopic dermatitis
Allergies

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
Mild-severe learning disability
Hyperactivity
Short attention span
Shy
Speech and language delay
High pitched voice or cry

CONTACT:
Contact a Family
Tottenham Court Road
London
W1P 0HA
Tel: 020 73833555
# Duplication 4p syndrome

## Alternative Names:

- [List of alternative names if available]

## General Health Screening:

Monitoring and assessment of cardiac functioning

## Physical Phenotype:

- Microcephaly
- Low set ears
- Beaked nose
- Micrognathia
- Chonal stenosis
- Proptosis
- Atrial-septal defect
- Left inguinal hernia
- Clinodactyly

## Psychological/Behavioural Phenotype:

- Learning disability
- Seizures

## Contact:

**UNIQUE – Rare Chromosome Disorder Support Group**

P.O. Box 2189
Surrey
CR3 5GN

Tel: 01883 330766
Web: [www.rarechromo.org](http://www.rarechromo.org)
**Duplication 9p syndrome**

**frequent prenatal mortality**

**ALTERNATIVE NAMES:**
- Partial trisomy 9
- Rethore syndrome
- 9p+ syndrome
- 9p duplication syndrome
- chromosome 9p trisomy
- dup (9p) syndrome
- trisomy 9p

**GENERAL HEALTH SCREENING:**
- Monitoring and assessment of cardiac function
- ENT assessment
- Monitoring and treatment of ear infections
- Renal assessment

**PHYSICAL PHENOTYPE:**
- Microcephaly
- Micrognathia
- Malformed protruding ears
- Hypertelorism
- Acentric displacement of pupils
- Cleft lip and palate
- Funnel chest widely spaced nipples
- Small hands clinodactyly
- Lordeosis and scoliosis
- Nail hypoplasia
- Congenital heart defects
- Kidney abnormalities
- Hypotonia
- Recurrent ear infections
- Constipation
- Speech delay and difficulty
- Osteoporosis-teenage years
- Atrial Septal Defect
- Motor disability

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**
- Learning disability
- High pain threshold

**CONTACT:**
UNIQUE – Rare Chromosome Disorder Support Group
P.O. Box 2189
Surrey
CR3 5GN
Tel: 01883 330766
Web: [www.rarechromo.org](http://www.rarechromo.org)
**Duplication 10q syndrome**
(most die in the first few years of life)

**ALTERNATIVE NAMES:**
- Chromosome 10q duplication syndrome
- 10q duplication syndrome
- Chromosome 10q trisomy
- Dup (10q) syndrome
- Partial trisomy 10q
- Trisomy 10q

**GENERAL HEALTH SCREENING:**
- Ophthalmic assessment
- Monitoring and assessment of cardiac function
- Genito-urinary assessment
- Renal assessment

**PHYSICAL PHENOTYPE:**
- Microcephaly
- Midfacial hypoplasia with flat facies
- Dolichocephaly
- Prominent cheeks
- High forehead flat round face
- Low set posteriorly rotated ears
- Microphthalmia
- Blepharophimosis
- Sclerocornea coloboma
- Short nose with a flat bridge
- Cleft lip or palate
- Camptodactyly, syndactyly, clinodactyly
- Scoliosis/kayphosis
- Hypotonia
- Heart defects
- Cryptorchidism
- Kidney aplasia or hypoplasia

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**
- Learning disability

**CONTACT:**
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- Surrey
- CR3 5GN
- Tel: 01883 330766
- Web: [www.rarechromo.org](http://www.rarechromo.org)
## Duplication 15q syndrome

(symptoms have been similar to those with Prader-Willi and Angelmans Syndrome)

### ALTERNATIVE NAMES:

- 15q+ syndrome
- 15q duplication syndrome
- chromosome 15q trisomy
- Dup (15q) syndrome
- Partial trisomy 15q syndrome
- Trisomy 15q

### GENERAL HEALTH SCREENING:

- Physiotherapy
- Monitoring and assessment of cardiac function
- Diabetic/thyroid assessment and screening
- Dietetics assessment
- Genito-urinary assessment

### PHYSICAL PHENOTYPE:

- Facial asymmetry
- Cloverleaf skull microdolichocephaly
- Microcephaly
- Micrognathia
- Hydrocephaly brain cysts
- Low set malformed ears
- Strabisms
- Cataracts
- Microphthalmia
- Prominent bulbous nose
- Cleft lip and palate
- Highly arched palate
- Inguinal and diaphragmatic hernia
- Camptodactyly
- Rocker bottom feet
- Joint and laxity contractures
- Scoliosis and kyphosis
- Café-au-lait patches
- Congenital heart defects
- Imperforate anus/jejunal artesia
- Cryptorchidism hypospadias amenorrhea absence or uterus horseshoe kidney and kidney reflux
- Diabetes mellitus and hypothyroidism
- Congenital heart defects

### PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

- Learning disability
- Speech delay aggressive and antisocial behaviour
- Obesity

### CONTACT:

Contact a Family
Tottenham Court Road
London
W1P 0HA

Tel: 020 73833555
## Dyggve-melchoir-classen syndrome

### ALTERNATIVE NAMES:
- DMC disease
- DMC syndrome
- Smith-McCort Dwarfism (this form does not have a learning disability)

### GENERAL HEALTH SCREENING:
- Physiotherapy

### PHYSICAL PHENOTYPE:
- Short stature
- Bulging of the chest sternum
- Flattening of the vertebrae and upper border of the pelvis (Iliac crest)
- Shortening of the metacarpals

### PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
- Learning Disability
Edwards Syndrome

ALTERNATIVE NAMES:

Trisomy 18: 18+ syndrome
3 forms of the syndrome:
- Full form (severe) every cell in the body has three no.18 chromosomes instead of two.
- Mosaic form (less severe) some cells have two no.18 chromosomes while others have three.
  The extent and severity of the condition will depend upon the ratio of normal to abnormal cells
- Partial form. Severity of the symptoms will depend upon whether the long arm (18q+) of the
  chromosome is affected Partial trisomy of the short arm clinical features may be absent and the
  learning disability may be mild or absent.

GENERAL HEALTH SCREENING:

Monitoring of hearing and vision
Assessment of continence
Dietetic assessment
Assessment and monitoring of cardiac and renal function
Monitoring and treatment of urinary tract infections

PHYSICAL PHENOTYPE:

Growth deficiency
Enlarged occiput
Small mouth and jaw
Cleft lip and palate
Small palpebral fissures
Microcephaly
Low set malformed ears
Clenched hands over lapping fingers
Bone abnormalities
Club/rocker-bottomed feet
Hypoplastic nails
Short sternum
Hernias
Skin mottling
Multiple congenital heart defects
Feeding and breathing problems(in infancy)
Oesophageal atresia
Diaphragmatic hernia
Hearing loss

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Learning Disability
Communication delay and limited to a few
words generally good non verbal
communication

CONTACT:

Contact a Family
Tottenham Court Road
London
W1P 0HA

Tel: 020 73833555
Web: www.cafamily.org.uk

Support organization for Trisomy 18,13 and
related disorders (SOFT)
Web: www.trisomy.org
# Fetal Warfarin Syndrome

## Alternative Names:
- Disala Syndrome
- Congenital Warfarin Syndrome
- Coumarin Syndrome
- Fetal anticoagulant Syndrome
- Heparin embryopathy
- Warfarin embryopathy
- Warfarin Syndrome

## General Health Screening:
- Ophthalmic assessment
- Hearing assessment
- Assessment and monitoring of cardiovascular function

## Physical Phenotype:
- Microcephaly
- Midface hypoplasia
- Optic atrophy
- Corneal opacity
- Cataracts
- Hypoplastic nose with low nasal bridge
- Choanal atresia
- Widely spaced nipples
- Shortening of the digits on the hands and feet
- Hypotonia
- Bone defects similar to those in chondrodysplasia punctata
- Puntata epiphyses
- Calcification disorders with stippling if the epiphyses
- Brain agesis
- Hydrocephalus
- Agenesis of the corpus callosum
- Meningoencephalocoele and seizures
- Patent ductus arteriosus
- Pulmonary stenosis
- Transposition of the great vessels
- Anomalous pulmonary veins
- Airway obstruction
- Deafness
- Feeding difficulties

## Psychological/Behavioural Phenotype:
- Learning Disability

## Contact:
- Contact a Family
- Tottenham Court Road
- London
- W1P 0HA
- Tel: 020 73833555
- Web: [www.cafamily.org.uk](http://www.cafamily.org.uk)
FG Syndrome

ALTERNATIVE NAMES:
Opitz-Kaveggia Syndrome

GENERAL HEALTH SCREENING:
Assessment of cardiac functioning
Ophthalmic assessment
Neurological assessment

PHYSICAL PHENOTYPE:
Short stature
High prominent forehead
Abnormalities of hair whorls: cowlicks
Hypertelorism
Prominent/everted lower lip
Imporforate anus (absence of the anal opening)
or an abnormally placed anus
Constipation
Hypotonia
Macrocephaly (large head)
Deafness
Congenital cardaic anomaly
Broad thumbs and big toes
Characteristic fingerprint patterns
Agenesis/hypoplasia of corpus callosum
Pyloric stenosis
Seizures

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
Learning disability
Hyperactive
Short attention span
Temper problems
Aggressive behaviour

Contact:
Contact a Family
Tottenham Court Road
London
W1P 0HA
Tel: 020 73833555
Web: www.cafamily.org.uk
### Floating Harbor Syndrome

**Alternative Names:**
- Pelletier-Leisti Syndrome

**General Health Screening:**
- Assessment and monitoring of cardiac function
- Dietetic assessment
- Dermatology assessment

**Physical Phenotype:**
- Short stature with delayed bone development
- Triangular face
- Prominent nose
- Deep set eyes
- Thin lips
- Short neck
- Finger abnormalities: brachydactyly and clinodactyly - clubbed end of fingers may be associated
- Seborrheic rash
- Hirtuism
- Cardiac septal defect
- Celiac disease

**Psychological/Behavioural Phenotype:**
- Learning Disability
- Delay in expressive language

**Contact:**
- Floating Harbour Support Group
  - 9 Crown Hill
  - Seaford
  - BN25 2XJ
  - Tel: 01323 891676

  or

  Contact a family: [www.cafamily.org.uk](http://www.cafamily.org.uk)
# Foetal alcohol syndrome

## Alternative Names:

- 

## General Health Screening:

- Cardiac Monitoring
- Hearing assessment
- Ophthalmic assessment

## Physical Phenotype:

- Growth retardation: head, weight and height
- Microcephaly
- Small maxilla (Jaw)
- Short upturned nose - sooth philtrum
- Smooth and thin upper lip narrow, small and unusual appearing eyes with prominent epicanthal folds
- Cardiac abnormalities: ventricular septal defect, atrial septal defect
- Limb abnormalities of joints, hands, feet, fingers and toes.
- Recurrent infections, mostly connected with the respiratory tract

## Psychological/Behavioural Phenotype:

### Contact:

- Fetal Alcohol Syndrome Trust
- PO Box 30
- Walton
- Liverpool
- L9 8HU

- Tel: 0151 284 2900

- [www.medcouncilalcol.demon.co.uk](http://www.medcouncilalcol.demon.co.uk)
# Fragile X Syndrome

## Alternative Names:
- Fra (X) Syndrome
- Martin Bell Syndrome
- Repenning Syndrome

## General Health Screening:
- Assessment and monitoring of cardiac function
- Orthodontic assessment
- Assessment and monitoring of epilepsy

## Physical Phenotype:
- A triad of physical features is evident: features are not always present in prepubertal males. In female carriers the features are more subtle.
- Large or prominent ears
- A long thin face with prominent jaw
- High arched palate, dental crowding and abnormal dermatoglyphics on hands and feet.
- Post pubertal testicular enlargement (often only affecting one testicle)
- Fingers can be easily hyperextended
- Flat feet
- Skin is often fine and thin
- Mitral valve prolapse
- Microcephaly
- Epilepsy
- Widespread central nervous system dysfunction

## Psychological/Behavioural Phenotype:
- Learning Disability
- Speech often delayed
- Gross fine motor co-ordination and motor planning remain poor
- Hyperactivity
- Concentration problems
- Irritability tantrums and aggressive outbursts
- Obsessive-compulsive disorders
- Lack of eye contact
- Stereotypical behaviours: hand flapping, resistance to environmental change, shy or anxious in social situations
- Easily overwhelmed by excessive sensory stimuli

## Contact:
- Fragile X Society
- 53 Winchelsea Lane
- Hastings
- East Sussex, TN35 4LG
- Tel: 014240813147
- [www.fragilex.org.uk](http://www.fragilex.org.uk)
- email: info@fragilex.org.uk
# Fraser Syndrome

## Alternative Names:

- Cryptohthalmos-syndactly syndrome
- Cryptohthalmos syndrome
- Cyclopism

## General Health Screening:

- Renal ultrasound and monitoring
- Genito-urinary assessment
- Ophthalmic assessment
- Audiology assessment and monitoring

## Physical Phenotype:

- Partial syndactly of fingers and toes
- Renal abnormalities dysplasia, hypoplasia, unilateral or bilateral agenesis
- Malformation of the eyes- blindness
- Cryptophthalmos (complete fusion of the eyelids)
- Malformations of the middle outer ear
- Skeletal anomalies
- Genital abnormalities:
  - Cryptorchidism one or both
  - Meatus (urinary opening) may be abnormally place on the underside of the penis or micro penis
  - Possible malformed fallopian tubes or clitoromegaly (an abnormally enlarged clitoris)
  - or bicornate uterus (an abnormally shaped uterus)
- The folds of skin on either side of the labia may be abnormally fused
- Anal atresia/stenosis
- Anophthalmia

## Psychological/Behavioural Phenotype:

- Learning Disability

## Contact:

FACES: The National Cranio Facial Association
USA
[http://www.faces-cranio.org](http://www.faces-cranio.org)

Tel: 423 266 1632
Galactosaemia

ALTERNATIVE NAMES:

GENERAL HEALTH SCREENING:
Assessment and monitoring of epilepsy
Dietetic assessment – Life long dietary treatment
Urology assessment
Assessment and monitoring of renal function
Counselling for fertility problems

PHYSICAL PHENOTYPE:
Cataracts
Liver disease
Late onset or cessation of puberty
Fertility is either reduced or infertile
Osteoporosis - in older people
Epilepsy
Present in first weeks of birth are,
  • feeding difficulties
  • vomiting
  • jaundice
  • liver and kidney disease

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
Speech delay and difficulties - oromotor dyspraxia
Learning difficulties specific to maths and science
May have a borderline learning disability
Visual perceptual difficulties
Anxiety
Social withdrawal

Contact:
Galactosaemia Support Group
31 Cotysmore Road
Sutton Coldfield
B75 6BJ

Tel: 0121 3785143
Email: Sue:gsg1.freeserve.co.uk
http://gsgnews.tripod.com
Goldenhar Syndrome

ALTERNATIVE NAMES:
- Facio-Auriculo-Vertebral Spectrum
- FAV
- Goldenhar-Gorlin Syndrome
- OAV Spectrum
- Occulo-Auriculo-Vertebral Dysplasia

GENERAL HEALTH SCREENING:
- Hearing assessment and monitoring
- Ophthalmic assessment and monitoring
- ENT assessment and monitoring
- Monitoring and assessment of cardiac function
- Monitoring and assessment of renal function

PHYSICAL PHENOTYPE:
- Asymmetry of the face affecting cheekbones, jaws, mouth, ears, eyes and/or bones of the spinal column
- Cleft lip
- Cleft palate
- Anotia (absence) and malformation or the auricles (outer ears)
- Narrow blind ending or absent external ear canals
- Preauricular Tags
- Abnormalities affecting the middle or inner ears-contributing or resulting in hearing loss
- Eye abnormalities
- Strabismus
- Hypoplasia, fusion and/or absence of certain vertebrae
- Cardiac abnormalities
- Pulmonary, renal and gastrointestinal abnormalities
- Obstructive sleep apnoea

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
- Mild learning disabilities in 5-15% of cases
- Emotional problems – due to facial disfigurement

Contact:
- Goldenhar Syndrome Hemifacial Microsomia
  Family Contact Group
  9 Harley Court Gardens
  Cranbrook
  TN17 3QY
  Tel: 01580 714042

or

Contact a Family
  www.cafamily.org.uk
**Hunter's Syndrome**

**ALTERNATIVE NAMES:**
Mucopolysaccharidosis tey 2  
Iduronate Sulfatase Deficiency  
Information below is for MPSIIA which is the severe form -life expectancy 10-15 years  
MPSIIB is the4 milder form -some of the characteristics are as severe but progress at a slower rate -life expectancy 50-60 years

**GENERAL HEALTH SCREENING:**
Assessment and monitoring of epilepsy  
Assessment and monitoring cardiac function  
ENT assessment  
Ophthalmic assessment  
Hearing assessment  
Liver function and cancer screening

<table>
<thead>
<tr>
<th>PHYSICAL PHENOTYPE:</th>
<th>PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:</th>
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<tbody>
<tr>
<td>Hydrocephalus</td>
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<td>Short stature</td>
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<td>Mild kyphosis</td>
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<tr>
<td>Upper airway obstruction</td>
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<td>Recurrent rhinorrhoea obstructive airway disease</td>
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<tr>
<td>Pulmonary hypertension</td>
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<tr>
<td>Cardiac functioning: valvular dysfunction, myocardial thickening, coronary artery narrowing, myocardial infarction</td>
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<tr>
<td>Chronic diarrhoea</td>
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<td>Inguinal/umbilical hernias</td>
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<tr>
<td>Severe retinal deterioration &amp; pigmentation</td>
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<tr>
<td>Recurrent otis media -progressive hearing loss</td>
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<td>Nodules of growth over the skin of the shoulders, scapulas, posterior chest wall and arm</td>
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<tr>
<td>Epilepsy</td>
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</tr>
</tbody>
</table>

**Contact:**
International Rare Disease Support Network  
[www.raredisorders.com](http://www.raredisorders.com)  
or information at,  
# Hurlers Syndrome

(Life expectancy approx. 10 years)

## ALTERNATIVE NAMES:

- Mucopolysaccharidosis type I
- Alpha-L-Iduronate deficiency (formerly gargoylism)
- Scheie (MPS IS) Syndrome
- Hurler/Scheie (MPS IH/S) syndrome

## GENERAL HEALTH SCREENING:

- Assessment and monitoring of cardiac function
- Assessment and monitoring of respiratory function
- ENT assessment

## PHYSICAL PHENOTYPE:

- Enlarged liver and spleen
- Corneal clouding
- Stiff joints
- Severe skeletal deformities
- Dwarfism
- Coarse facial features
- Enlarged tongue
- Prominent forehead
- Hearing loss
- Visual loss
- Hirsutism
- Recurrent respiratory infections
- Otis media
- Upper airway obstruction
- Heart disease
- Persistent rhinorrhea
- Obstructive hydrocephalus
- Kyphosis
- Umbilical and inguinal hernias

## PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

- Severe Learning Disability

## Contact:

The Society for Mucopolysaccharide Diseases
46 Woodside Road
Amersham
Buckinghamshire
HP6 6AJ

Tel: 01494 434156

Email: mps@mpssociety.co.uk
Web: [www.mpssociety.co.uk](http://www.mpssociety.co.uk)
[www.fabry.org.uk](http://www.fabry.org.uk)
Johanson Blizzard Syndrome

ALTERNATIVE NAMES:

JBS
Nasal Alar Hypoplasia, Hypothyroidism, Pancreatic Achylia, Cong. Deafness
Ectodermal Dysplasia - Exocrine Pancreatic Insufficiency
Malabsorption - Ectodermal Dysplasia - Nasal Hypoplasia

GENERAL HEALTH SCREENING:

Assessment and monitoring of cardiac function
Monitoring and treatment of hypothyroidism
Hearing assessment
Dietetic assessment
Glucose Tolerance tests and Sweat tests
Assessment and monitoring of diabetes

PHYSICAL PHENOTYPE:

Small nose-aplasia or hypoplasia of the nostrils
Abnormally formed deciduous teeth and misshapen or absent permanent teeth
Unusually sparse, dry, coarse scalp with distinctive upsweep in the forehead
Abnormal development of the pancreas
Short stature
Hypothyroidism
Hearing loss
Speech problems
Microcephaly
Cardiac problems; situs inversus/ dextrocardia, atrial septal defect
Poor muscle tone
Imperforated anus
Greater susceptibility to respiratory infections

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Mild to Severe Learning Disability

Contact:
Self Help Group
RTMDC (Research Trust for Metabolic Diseases in Children)
Weston Road
Crewe
Cheshire
CW1 1XN
Tel: 01270 250221
Pallister (PKS) Killian Syndrome

ALTERNATIVE NAMES:
Pallister-Killian Syndrome
Chromosome 12
Isochromosome 12p mosaic
Killian Syndrome
Pallister Mosaic Syndrome
Pallister Mosaic Syndrome Tetrasomy 12P

GENERAL HEALTH SCREENING:
Monitoring and treatment of epilepsy
Ophthalmic assessment
Monitoring and assessment of cardiac function
ENT assessment
Monitoring and treatment of hernia

PHYSICAL PHENOTYPE:
Coarse face with high forehead
Sparse hair on scalp
Hypertelorism
Epicanthal folds
Strabismus
Ptosis
Broad nasal bridge with high arched palate
Long philtrum with thin upper lip with cupid bow shape
Protruding lower lip-delayed dental eruption
Large ears with thick protruding lobules
Hypotonia
Sparse hypopigmented macules
Hearing loss
Accessory nipples
Laryngomalacia
Gastroesophageal reflux
Cataracts
Congenital heart defects
Diaphragmatic hernia
Epilepsy
Hypopigmentation
Hyperpigmentation

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
Learning Disability
Minimal speech development

Contact:
UNIQUE
Rare Chromosome Disorder Support Group
PO Box 2189
Caterham
CR3 5GN
Tel: 01883 330766
Email: info@rarechromo.org
Web: www.rarechromo.org
# Langer-Giedion Syndrome

**ALTERNATIVE NAMES:**

Trichlorhinophalangeal Syndrome type II (TRPS2)
(this is a contiguous Syndrome)

**GENERAL HEALTH SCREENING:**

Physiotherapy
Hearing assessment
Assessment and monitoring of respiratory function

**PHYSICAL PHENOTYPE:**

- Cranio facial abnormalities
- Large, laterally protruding ears
- Broad nasal bridge and bulbous nose
- Elongated upper lip, with thin vermillion border
- Broad eye brows
- Sparse hair
- Mild microcephaly
- Multiple cartilaginous exostoses
- Cone shaped epiphyses
- Possibly: short stature, overly flexible joints
- Excess fold of skin

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**

Possible Learning Disability
## Lawrence-Moon-Biedl Syndrome
(Differing articles claim connection and separation to the Bardet-Biedl Syndrome. Both highlight Learning Disability)

### ALTERNATIVE NAMES:
- Laurence Moon Syndrome
- Bardet-Biedl Syndrome
- Adipogential-Retinitis Pigmentosa Syndrome
- Laurence Syndrome
- LM Syndrome

### GENERAL HEALTH SCREENING:
- Ophthalmic assessment
- Endocrinology assessment
- Renal and hepatic assessment
- Monitoring and assessment of diabetes
- Dietetic assessment

### PHYSICAL PHENOTYPE:
- Hypogonadism
- Retinitis pigmentosa (progressive loss of vision)
- Spastic paraplegia
- Obesity
- Polydactyly
- Kidney malformations and renal dysfunction
- Speech difficulties
- Diabetes mellitus
- Hepatic fibrosis
- Hormonal deficiencies

### PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
- Learning Disability

### Contact:
- LMBB Society
  1 Blackthorn Avenue
  South Borough
  Tunbridge Wells
  TN4 9YA
- Tel: 01892 682680
- Email: Julie.sales@lmbbs.org.uk
- Web: [www.lmbbs.org.uk](http://www.lmbbs.org.uk)
# Marden Walker Syndrome

**ALTERNATIVE NAMES:**

Connective Tissue Disorder, Marden walker type  
MWS

**GENERAL HEALTH SCREENING:**

Assessment and monitoring of cardiac function  
Hearing assessment

**PHYSICAL PHENOTYPE:**

| Cleft or high arched palate  
| Bone joints in a fixed position  
| Growth delay  
| Limited control of muscle movement  
| Pectus excavatum  
| Arachnodactyly, camptodactyly, talipes equinovarus and transverse palmer creases  
| Scoliosis and kyphosis  
| Atrial septal and ventricular septal defects and cardiomyopathy  
| Low set ears/malformed  
| Upturned nose  
| Eyes:- blepharoshimosis, extropia/esotropia and Blepharoptosis  
| Multiple Contractures :- elbow, knee and hip joints  
| Nervous System:- Absent Moro and deep tendon reflexes

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**

Learning Disability

**Contact:**

Contact a Family  
Tottenham Court Road  
London  
W1P 0HA  
Tel: 020 73833555  
Web: [www.cafamily.org.uk](http://www.cafamily.org.uk)
Marshall Smith Syndrome

ALTERNATIVE NAMES:

GENERAL HEALTH SCREENING:
Monitoring and assessment of respiratory function
Monitoring and assessment of weight
Dietetics assessment
Ophthalmic assessment
Hearing assessment
Monitoring and assessment of skeletal changes

PHYSICAL PHENOTYPE:
Accelerated skeletal maturation
High birth weight but failure to thrive
Congenital cardiac anomalies
Macrocephaly
Myopathy
Optic atrophy
Hypertelorism
Palapable fissure slant down
Hirsuism
Facial anomalies-thin lower lip/upper lip, prominent eyes
Sclerosis/thick skull
Hydronephrosis
Narrow thorax
Narrow atretic auditory canal
Osteosclerosis or osteoporosis
Treachea or laryngeal anomalies

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
Learning Disability
# Mucopolysaccaridosis VII

**ALTERNATIVE NAMES:**

- Sly syndrome
- Beta-glucuronidase Deficiency
- Beta-glucuronidase deficiency mucopolysaccaridosis
- GUSB deficiency
- Mucopolysaccaride storage disease VII

**GENERAL HEALTH SCREENING:**

- Ophthalmic assessment
- Monitoring and treatment of respiratory infection
- Monitoring and assessment of renal function

**PHYSICAL PHENOTYPE:**

- Moderate skeletal abnormalities
- Inguinal/umbilical hernias
- Hepatosplenomegaly
- Coarse facies
- Short stature
- Recurrent respiratory infections
- Hypertelorism, corneal opacity and iris colobomata
- Prominent alveolar processes and cleft palate
- Hepatomegaly
- Slenomegaly

There is a wide spectrum of severity spanning from severe psychomotor delay and progressive disease leading to death in the first years of life to a normal phenotype in the second decade

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**

- Growth, motor and learning disability

**Contact:**

The Society for Mucopolysaccharide Diseases
46 Woodside Road
Amersham
HP6 6AJ

Tel: 01494 434156
Email: mps@mpssociety.co.uk
Web: www.mpssociety.co.uk
www.fabry.org.uk
Opitz Syndrome

**ALTERNATIVE NAMES:**
- Opitz-Frias Syndrome
- Autosomal Dominant Opitz Syndrome
- BBB Syndrome
- BBBG Syndrome
- BBB/G Syndrome
- BBG Syndrome
- G Syndrome
- GBBB Syndrome
- G/BBB Syndrome
- Hypospadias-Dysphagia Syndrome
- Oculo-Genito-Laryngeal Syndrome
- Telechanthus-Hypospadias Syndrome
- X-Linked Opitz Syndrome

**GENERAL HEALTH SCREENING:**
- Monitoring and assessment of cardiac function
- Speech and Language Therapy (assessment of swallow reflex)
- Dietetics assessment
- Monitoring and assessment of renal function
- Hearing assessment

**PHYSICAL PHENOTYPE:**
- Hypertelorism
- Ears low set or rotated at an unusual angle
- Broad/flat nose
- Small chin
- Hypospadias
- Craniofacial anomalies
- Congenital heart defects
- Laryngotraheal Disorders with dysphagia and aspiration
- Hernias- inguinal
- Kidney abnormalities
- Cleft lip
- Laryngeal cleft
- Horse cry
- Cryptarchidism
- Bitid Scrotum
- Imperforated anus

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**
- Mild learning disabilities in some cases

**Contact:**
- Contact a Family
  - Tottenham Court Road
  - London
  - W1P 0HA
  - Tel: 020 73833555
  - Web: [www.cafamily.org.uk](http://www.cafamily.org.uk)
# Phenyketonuria
(treatable following diagnosis with Guthrie test)

## ALTERNATIVE NAMES:

Information provided is reflective of PKU if untreated. However ( ) information is reflective of MPKU
Maternal PKU syndrome (irreversible)

## GENERAL HEALTH SCREENING:

Dietetic assessment
Assessment and monitoring of cardiac functioning
Assessment and monitoring of epilepsy
Assessment and monitoring of mental health
Periodic developmental screening
Biochemical monitoring
Neurological assessments

## PHYSICAL PHENOTYPE:

- Usually fair skinned with blonde hair and blue eyes or fairer complexion than family members (due to phenylalanine's role as precursor of melanin)
- Inability to process the amino acid phenylalanine
- Microcephaly (MPKU also)
- Congenital heart defects (MPKU also)
- Low birth weight (MPKU only)
- Poor co0rdination
- Dystonia, Athetoid movements
- Ataxia
- Unusual odour detected on breath, skin, urine from the accumulation of phenylacetic acid
- Eczema
- Facial dysmorphism (MPKU only)
- Premature closure of cranial sutures (MPKU only)
- Epilepsy

## PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

- Learning Disability (MPKU also)
- Hyperactivity
- Agrophobia
- Behavioural difficulties: aggression, self inflicted injuries, impulsivity, psychosis
- Attention Deficit and Hyperactivity Disorder (ADHD) (MPKU only)
- Mental health problems including depression (MPKU only)
- Childhood autism (MPKU only)
- Anxiety
- Delayed mental and social skills

## Contact:

Contact a Family
Tottenham Court Road
London
W1P 0HA
Tel: 020 73833555 Web: [www.cafamily.org.uk](http://www.cafamily.org.uk)
or
National Society for Phenylketonuria (UK)
Ltd, London
Tel: 020 83643010
Peter's Plus Syndrome

**ALTERNATIVE NAMES:**

Krause-Kivlin Syndrome
Peters Anomaly-short limb dwarfism syndrome

**GENERAL HEALTH SCREENING:**

Cardiac assessment and monitoring
Ophthalmology assessment
Audiology assessment
ENT assessment
Assessment and monitoring for epilepsy
Genito-urinary assessment

**PHYSICAL PHENOTYPE:**

**Short stature**
Short fingers and toes
Cleft lip and palate (not always present)
Malformed and prominent ears
Peters anomaly – defects on the anterior chamber of the eye, including central corneal opacity. Thinning of the posterior aspect of the cornea and adhesions of the iris, lens or cornea.

Narrow eye lid closure
Cardiovascular abnormalities
Motor and growth retardation
Failure to thrive
Urogenital abnormalities
Agenesis of the corpus callosum (not always present)

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**

Mild to severe learning disabilities (not always present)
Epilepsy
# Prader-Willi Syndrome

## Alternative Names:
- PWS

## General Health Screening:
- Dietetic assessment and nutritional advice
- Assessment and monitoring of cardiac function
- Psychology assessment
- Assessment and monitoring of epilepsy
- Ophthalmic assessment
- Regular checks on blood/urine glucose levels

## Physical Phenotype:
- Short stature
- Almond shaped eyes
- Narrow-difrontal diameter
- Triangular mouth
- Early dental caries/enamel hypoplasia
- Hypogonadism/cryptorchidism
- Insatiable appetite
- Reduced calorie intake requirement
- Obesity
- Hypertension
- Respiratory difficulties
- Diabetes mellitus
- Hypotonia (in childhood)
- Small hands and feet
- Co-ordination and balance difficulties
- Sleep apnoea
- Epilepsy

## Psychological/Behavioural Phenotype:
- Learning Disability
- Behavioural problem: temper outbursts, stubborness, rigidity, argumentativeness, repetitive thoughts and behaviours. Skin picking
- Speech and language difficulties - expressive skills often being considered poorer than receptive skills

## Contacts:
- Prader-Willi Syndrome Association (UK)
  - 125a London Road
  - Derbe
  - DE1 2QQ
  - Tel: 01332 365676
  - Web: [www.pwsa-uk.demon.co.uk](http://www.pwsa-uk.demon.co.uk)
## Rett Syndrome

### ALTERNATIVE NAMES:
- RS
- RTS

### GENERAL HEALTH SCREENING:
- Speech and language therapy assessment (swallowing)
- Assessment and monitoring for epilepsy
- Orthotic assessment
- Podiatrist assessment
- Dietetic assessment

### PHYSICAL PHENOTYPE:
**Occurs mainly in females although can on rare occasions occur in males**
- Scoliosis
- Rigidity
- Dystonia
- Foot deformities/ cold feet
- Breathing problems
- Failure to thrive (not always present)
- Gastrointestinal difficulties – chronic constipation
- Growth retardation
  - In adulthood most likely to be in stage iii or stage iv A/B
- Stage iii – increased motor difficulties and epileptic seizures
- Stage iii (A previously ambulant) (B never ambulant) – reduced mobility, no further decline in communication or hand skills. Increased rigidity and dystonia.
- Slow development from birth
- Bruxism
- Facial movements

### PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:
- Mild to severe learning disabilities
- Epilepsy
- Mood fluctuation
- Social withdrawal
- Self injurious behaviour in some

### Contact:
- Rett Syndrome Association UK
  - Web: [www.rettsyndrome.org.uk/](http://www.rettsyndrome.org.uk/)
- Or
  - Contact a Family
  - Tottenham Court Road
  - London
  - W1P 0HA
  - Tel: 020 73833555
  - Web: [www.cafamily.org.uk](http://www.cafamily.org.uk)
# San Fillippo Syndrome

**ALTERNATIVE NAMES:**

The four sub-types of this syndrome are difficult to distinguish clinically by they differ biochemically. A, B, C, D, (MPS IIIA, IIIB, IIIC, IIID)

**GENERAL HEALTH SCREENING:**

- Monitoring and treatment of respiratory infections
- ENT assessment
- Dietetic assessment
- Assessment and monitoring of epilepsy
- Hearing assessment

**PHYSICAL PHENOTYPE:**

- Severe progressive CNS involvement
- Hirsutism
- Hepatosplenomegaly
- Severe hearing loss
- Recurrent ear, nose and throat infections
- Episodes of diarrhoea are common
- Coarse facial features develop later in this condition
- Neurological degeneration
- Balance is affected and falls are common
- Aspiration is noted with feeding difficulties most will require tube feeding
- Spasticity and joint stiffness deterioration leads to impaired mobility, requiring wheelchair support
- Epilepsy - tonic clonic seizures

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**

- Learning Disability
- Hyperactivity
- Behavioural problems
- Speech is usually apparent then gradually lost

**Contact:**

Society for Mucopolysaccharide diseases
46 Woodside Road
Amersham
Buckinghamshire
HP6 6AJ

Tel: 01494 434156

Or

Email: mps@mpssociety.co.uk
Web: www.mpssociety.co.uk
www.fabry.org.uk
# Smith-Lemli-Opitz Syndrome

## Alternative Names:
- Smith-Lemli-Opitz/RSH Syndrome
- SLO/RSH
- Smith Syndrome
- RSH/Smith-Lemli-Opitz (RSG/SLO) syndrome

## General Health Screening:
- Dietetic assessment
- Assessment and monitoring of cardiac function
- Renal assessment
- Cholesterol testing
- Surgical referral re: Pyloric stenosis

## Physical Phenotype:
- Cleft lip and palate
- Polydactyly
- Microcephaly
- Low set ears
- Small upturned nose
- Webbing between 2nd and 3rd toes
- Abnormal palmar creases (usually single)
- Hypospadias
- Undecended testes
- Cataracts
- Belpharoptosis
- Heart defects eg. Ventricular dilation
- Micrognathia
- Pyloric stenosis
- Hirschprung disease (absent nerves in colon)
- Abnormal metabolism or cholesterol
- Eyes- strabismus, cataracts
- Tongue abnormalities
- Hypoplasia
- Blond hair
- Brain abnormalities
- Genital abnormalities
- Recurrent infections
- Feeding difficulties
- Renal defects

## Psychological/Behavioural Phenotype:
- Learning Disability
- Abnormal sleep pattern
- Photosensitivity
# Smith-Magenis Syndrome

## Alternative Names:

Interstitial deletion of chromosome 17

## General Health Screening:

- Hearing assessment
- Ophthalmic assessment
- Monitoring and treatment of ear infections
- Monitoring of cardiac function
- Monitoring for possible seizures

## Physical Phenotype:

- Short stature
- Flat facial features
- Prominent jaw
- Down turned mouth
- Unusually formed ears
- Chronic ear infections
- Hearing impairments
- Eye problems including: strabismus, nearsightedness
- Short fingers and toes
- Scoliosis
- Unusual gait
- Less frequently - Heart defects and murmurs
- Urinary system problems
- Abnormalities of the palate, with or without cleft lip

## Psychological/Behavioural Phenotype:

- Learning disability
- Behaviour problems
- Sleep problems
- Self injurious behaviour

## Contact:

**Smith-Magenis Support Group**
1 Poppyfields
Chester-le-Street
County Durham
DH2 2NA
Tel: 0191 3888868

Or

**Smith-Magensis Syndrome Foundation**
81 Cedar Ridge
Dungannon
BT71 6UD
Tel: 02887 750050

Email: gmc@yolger.fsnet.co.uk
# Soto's Syndrome

**ALTERNATIVE NAMES:**

- Cerebral gigantism

**GENERAL HEALTH SCREENING:**

- Assessment and monitoring of epilepsy
- Treatment and management of constipation
- Monitoring and treatment of respiratory infections
- Dietetic assessment
- Behavioural assessment - psychology support

**PHYSICAL PHENOTYPE:**

<table>
<thead>
<tr>
<th>Physical Features</th>
<th>Psychological/Behavioural Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>Facial changes are evident with age from rounded in infancy to long and thin with age</td>
<td>Mild to severe Learning Disability</td>
</tr>
<tr>
<td>Hypotonia may persist</td>
<td>Aggressive behaviour</td>
</tr>
<tr>
<td>Joint laxity and pes planus</td>
<td>Ritualistic behaviour</td>
</tr>
<tr>
<td>Speech impairment</td>
<td>Sleeping problems</td>
</tr>
<tr>
<td>Macrocrania</td>
<td>Attention deficits</td>
</tr>
<tr>
<td>Disproportionately large head with a slightly protrusive forehead</td>
<td>Hyperactivity</td>
</tr>
<tr>
<td>Large hands and feet</td>
<td>Contacts:</td>
</tr>
<tr>
<td>High arched palate</td>
<td>Soto’s Group Child growth Foundation</td>
</tr>
<tr>
<td>Premature eruption of teeth</td>
<td>2 Mayfield Road</td>
</tr>
<tr>
<td>Sparseness of hair</td>
<td>Chiswick</td>
</tr>
<tr>
<td>Hypertelorism</td>
<td>W4 1PW</td>
</tr>
<tr>
<td>Downslanting eyes</td>
<td>Tel: 0181 9950257</td>
</tr>
<tr>
<td>Clumsiness, awkward gait</td>
<td>Or</td>
</tr>
<tr>
<td>Constipation</td>
<td>Soto’s Syndrome Support Association</td>
</tr>
<tr>
<td>Persistent drooling and reluctance to chew</td>
<td>Email: <a href="mailto:sssa@well.com">sssa@well.com</a></td>
</tr>
<tr>
<td>Puberty: Females - early menarche. Males delayed puberty</td>
<td>Web: <a href="http://www.well.com/user/sssa/">www.well.com/user/sssa/</a></td>
</tr>
<tr>
<td>Frequent respiratory infections - often resulting in conductive hearing loss</td>
<td></td>
</tr>
<tr>
<td>Cerebral: ventricular dilation</td>
<td></td>
</tr>
<tr>
<td>Seizures - possibly only febrile</td>
<td></td>
</tr>
<tr>
<td>Wilms Tumour – renal tumour</td>
<td></td>
</tr>
</tbody>
</table>
### Tuberous Sclerosis

**ALTERNATIVE NAMES:**

- Bournville’s Disease

**GENERAL HEALTH SCREENING:**

- Dermatology assessment and treatment
- Monitoring and management of epilepsy
- Monitor blood pressure
- Assessment of renal function
- Hearing assessment
- Ophthalmologic assessment
- Echocardiography screening
- Electrocardiograms (if abnormal heart rhythm)
- CTScan for lesions on lung
- Neurodevelopment testing

**PHYSICAL PHENOTYPE:**

- Epilepsy
- Skin discoloration, rashes.
- Calcified growths in the brain
- Cysts or angiomyolipomata (benign growths) in the kidneys
- Rhabdomyomata (benign growth of heart muscle
- Hearing difficulties
- Skin and eye lesions

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**

- Some degree of learning disability in 50% of cases
- Behaviour problems – neurobehavioral problems
- Autistic tendencies
- Hyperactivity, attention deficit, obsessive compulsive behaviours
- Occasional schizophrenia
- Bi polar disorder
- Depression
- Sever sleep disturbance

**Contact:**

- Tuberous sclerosis Alliance – [www.tsalliance.org](http://www.tsalliance.org)
- Contact a family – [www.cafamily.org.uk/direct/t45.html](http://www.cafamily.org.uk/direct/t45.html)
# Trisomy 8 Syndrome

**ALTERNATIVE NAMES:**
- Warkany Syndrome
- 8 Trisomy
- Trisomy 8

**GENERAL HEALTH SCREENING:**
- Ophthalmic assessment
- Assessment and monitoring of cardiac function
- Renal assessment
- Urology assessment
- Dietetic assessment

**PHYSICAL PHENOTYPE:**
- Congenital heart defects/septal defects
- Kidney malformation
- Everted thick lips
- High arched or cleft palate
- Prominent ears
- Broad upturned nose
- Stabismus
- Hypertelorism
- Corneal opacity
- Cataract
- Heterochromia
- Downslanting palpebral fissure
- Camptodacly
- Clinodacty
- Short metacarpal/tarsals other anomalies
- Absent or dysplastic patellae
- Multiple joint contractures
- Abnormal diaphyses and epiphyses of radial, femoral and humeral bones
- Kyphosis
- Spina bifida
- Uretal reflux and hydronephrosis
- Cryptorchidism
- Malrotation or absence of gall bladder

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**
- Learning Disability
- Speech delay

**Contact:**
- UNIQUE – Rare Chromosome Disorder Support Group
  - P.O. Box 2189
  - Surrey
  - CR3 5GN
  - Tel: 01883 330766
  - Web: [www.rarechromo.org](http://www.rarechromo.org)
# Trisomy 9 Mosaic Syndrome

**ALTERNATIVE NAMES:**
- Trisomy 9 / complete trisomy 9 syndrome
- Trisomy 9 mosaic
- Trisomy 9 mosaicism
- Trismoy 9 mosaicism syndrome

**GENERAL HEALTH SCREENING:**
- Assessment and monitoring of congenital heart defects
- Renal assessment

**PHYSICAL PHENOTYPE:**
- Growth deficiency
- Sloping forehead
- Bulbous nose
- Palpebral fissures
- Small deepset eyes
- Short eyelid folds
- Low set malformed ears
- Small jaw
- Large fontanelles
- Congenital heart defects
- Kidney abnormalities
- Skeletal abnormalities
- Musculoskeletal abnormalities
- Craniofacial abnormalities

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**
- Learning disabilities

**Contact:**
- UNIQUE – Rare Chromosome Disorder Support Group
- P.O. Box 2189
- Surrey
- CR3 5GN
- Tel: 01883 330766
- Web: [www.rarechromo.org](http://www.rarechromo.org)
- Email: [info@rarechromo.org](mailto:info@rarechromo.org)
## Turner Syndrome

### ALTERNATIVE NAMES:

Ullrick- Turner Syndrome

### GENERAL HEALTH SCREENING:

- Monitor and treatment of high blood pressure
- Monitor and treatment of hypothyroidism
- Hearing assessment and monitoring

### PHYSICAL PHENOTYPE:

- Short stature
- Osteoporosis
- Cubitus Valgus (increased carrying angle elbow)
- Short metacarpals (bone of fingers or Knuckles)
- High arched palate
- Infertility
- Webbed neck
- Low posterior hairline
- Swelling of hands and/or feet
- High blood pressure
- Structural abnormalities in kidneys
- Hypothyroidism
- Carbohydrate intolerance

### PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

- Normal range of intelligence
- Difficulties in concentration
- Overactive behaviours

### Contact:

Turner Syndrome Support Society (UK)
12 Irving Quadrant
Hardgate
Clydebank
G81 6AZ
Tel: 01389 380385

Email: Turner.syndrome@tss.org.uk
Web: www.tss.org.uk
Wolf-Hirschhorn Syndrome

**ALTERNATIVE NAMES:**
4p Deletion Syndrome
Monosomy 4p
wolf-Hirschhorn Syndrome 4p

**GENERAL HEALTH SCREENING:**
Monitoring and assessment of epilepsy
Hearing assessment
Ophthalmic assessment
Cardiac assessment
Monitoring and assessment respiratory function

**PHYSICAL PHENOTYPE:**
Facial dysmorphic features
Microcephaly
Midline scalp defect
Hemangioma of the forehead
Hypertelorism
Downward, slanting palpebral fissures
Epicanthus
Strabismus
Colobotoma
Low set simple ears
Preauricular dimples
Broad or beaked nose
Cleft lip, palate, and/or uvula
Carp-like mouth
Microganthia
Epilepsy
Hypospadias
Heart defects
Recurrent respiratory infections

**PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:**
Severe Learning Disability

**Contact:**
Mrs Chris Hilder
Wolf – Hirschhorn Syndrome Support Group
2b Harvesters Close
Rainham
Gillingham
ME8 8PA
Tel: 01634 264816
Email: whs@webk.co.uk
Web: www.whs.webk.co.uk
X-Linked Hydrocephalus

ALTERNATIVE NAMES:

MASA Syndrome
Bickers-Adams Syndrome
HSAS, HYCX
XLAS

GENERAL HEALTH SCREENING:

Ophthalmic assessment
Neurological assessment: VP shunt
Assessment of cardiac function

PHYSICAL PHENOTYPE:

Enlarged head
Large anterior fontanels, separated sutures
Organ anomalies…
Flexion deformity of the thumbs
Aqueductal stenosis
Chorioretinitis
Hydrocephaly/ large ventricles, non specific
Aphasia / shuffling gait

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Learning disability
Speech deficit

Contact:

Contact a Family
Tottenham Court Road
London
W1P 0HA

Tel: 020 73833555
Web: www.cafamily.org.uk
Useful Websites and Contacts

ACCESSIBLE INFORMATION

Change Picture Bank
www.changepeople.co.uk

Confederation of Transcribed Information Services (COTIS)
www.cotis.org.uk

Easy Information
www.easyinfo.org.uk

Real Voice Media
www.realvoice.org

Royal National Institute for the Blind Accessible Information
www.rnib.org.uk/access/

ADVOCACY AND SELF HELP ORGANISATIONS

Central England People First
www.peoplefirst.org.uk

Citizen Advocacy Information and Training
www.citizenadvocacy.org.uk

Disability Rights Commission
www.drc-gb.org

Disabled Peoples' International
Disabled Peoples' International is a network of national organizations or assemblies of disabled people, established to promote human rights of disabled people through full participation, equalization of opportunity and development.
www.dpi.org/

First Step (Merseyside)
Breckfield Centre
Breckfield Road North
Everton
Liverpool L5 0QW
Tel: 0151 222 0575
Contacts: Karen Flood & Bill Heron

Greater Manchester Coalition of Disabled People (GMCDP)
Information, advocacy, peer support, quarterly magazine, volunteering and organising opportunities and has a vibrant Young Disabled People's Forum which includes a drama group.
0161 273 5154
www.changepeople.co.uk
Confederation of Transcribed Information Services (OTIS)
www.cotis.org.uk

Easy Information
www.easyinfo.org.uk

Real Voice Media
www.realvoice.org

Royal National Institute for the Blind Accessible Information
www.rnib.uk/access/

ADVOCACY AND SELF HELP ORGANISATIONS

Central England People First
www.peoplefirst.org.uk

Citizen Advocacy Information and Training
www.citizenadvocacy.org.uk

Disability Rights Commission
www.drc-gb.org

Disabled Peoples’ International
Disabled Peoples’ International is a network of national organizations or assemblies of disabled people, established to promote human rights of disabled people through full participation, equalization of opportunity and development.
www.dpi.org/

First Step (Merseyside)
Breckfield Centre
Breckfield Road North
Everton
Liverpool L5 0QW
Contacts: Karen Flood & Bill Heron

Greater Manchester Coalition of Disabled People (MCDP)
Information, advocacy, peer support, quarterly magazine, volunteering and organising opportunities and has a vibrant Young Disabled People’s Forum which includes a drama group.
0161 273 5154
Business Employment Venture Centre, Aked Close, Ardwick, Manchester, M12 4AN
www.gmcdp.com
Heart and Soul  
www.heartnsoul.co.uk

One For Us  
www.oneforus.com

People First (National)  
www.peoplefirstltd.com

Plain Facts Magazine  
www.bris.ac.uk/Depts/NorahFry/PlainFacts/index.html  
Relate  
www.relate.org.uk

Samaritans  
www.samaritans.org.uk

Values Into Action  
www.viauk.org

ISSUES AROUND AGEING

Age Concern England  
www.ace.org.uk/

Ageing with Developmental Disabilities  
tigger.uic.edu/orgs/rrtcamr

Centre for Policy on Ageing  
www.cpa.org.uk

Counsel and Care  
www.counselandcare.org.uk

ANXIETY DISORDERS

Anxiety Community  
www.anxietyhelp.org

First Steps – Obsessive Compulsive Disorder  
www.first-steps.org

National Phobics Society  
www.phobics-society.org.uk

No Panic  
www.no-panic.co.uk

Triumph Over Phobia  
www.triumphoverphobia.com
ADHD Attention Deficit Hyperactive Disorder
Attention Deficit Disorder Association (USA)
www.add.org

AUTISTIC SPECTRUM DISORDERS

Asperger’s Syndrome
www.wpi.edu/~trek/aspergers.html

Asperger’s Syndrome
www.aspergerssyndrome.co.uk

For information and training about Asbergers Syndrome
Contact Michelle Evans – slinkyshellrules@yahoo.co.uk
For children contact Karen Penlington – kpenlington7@hotmail.com

Autism Connect
www.autismconnect.org

Autism Europe
www.autismeurope.arc.be

Autism Independent UK
www.autismuk.com

Autism in Mind
www.autism-in-mind.org

Autism Network International
www.ani/autistics.org

Autism Research Centre (University of Sunderland)
www.osiris.sunderland.ac.uk

Autism Society Of America
www.autism-society.org

Centre For The Study Of Autism
www.autism.org

Cumbria Support Group
www.southlakesautism.co.uk
autisticspectrumdisorders@postmaster.co.uk

National Autistic Society (UK)
www.nas.org.uk

Looking Up (Monthly International Autism Newsletter)
www.lookingupautism.org

The Maze: Autism Links & Information
www.isn.net/~jypsy/autilink.htm
World Autism  
www.worldautism.org

Yale University Centre For Autism And Developmental Disabilities  
www.info.med.yale.edu/chldstdy/autism

BRAIN INJURY

British Institute for Brain Injured Children  
www.bibic.org.uk

CARERS

Caring about Carers  
www.carers.gov.uk

Carer's UK  
www.carersonline.org.uk

Princess Royal Trust for Carers
Have a chat room where you can talk with other carers and a message board. People get together at 7pm on the internet every evening.  
Tel: 020 7480 7788  
www.carersonline.org

CEREBRAL PALSY

Scope  
www.scope.org.uk

CHALLENGING BEHAVIOUR

Challenging Behaviour Foundation  
www.thecbf.org.uk

Institute of Applied Behaviour Analysis  
www.iaba.net

CHILDREN’S ISSUES

Childline  
www.childline.org.uk

NSPCC  
www.nspcc.org.uk
Hyperactive Children’s Support Group  
www.hacsg.org.uk

International Association Of Infant Baby Massage  
www.iaim.org.uk

National Blind Children’s Society  
www.nbcs.org.uk

Scope’s Early Years website  
www.scope.org.uk/earlyyears/

SIBBS (Support In Bereavement For Brothers & Sisters)  
www.tcf.org.uk

CHILD AND ADOLESCENT MENTAL HEALTH

About Our Kids  
www.aboutourkids.org

Centre for Adolescent & Family Studies  
www.education.indiana.edu/cas/adol/mental.html

Child, Adolescent & Family Mental Health Research Centre  
www.worc.ac.uk/departs/hsc/CAMH.html

Young Minds  
www.youngminds.org.uk

COMMUNICATION

ACE Centre (Aids to Communication in Education)  
www.ace-centre.org.uk

British Stammering Association  
www.stammering.org

Communication Matters  
www.communicationmatters.org.uk

Breaking Barriers  
This site is one of the outcomes of meetings with alternative communication users.  
www.breaking-the-barriers.org/

AFASIC (Speech, Language and Communication)  
www.afasic.org.uk
Audit Commission have published a report into the role of assistive technology in supporting independence. www.audit-commission.gov.uk/reports/NATIONAL-REPORT.asp?CategoryID

Communications Forum
www.communicationsforum.co.uk

FACT
Bolton Institute
www.inclusion-boltondate.org.uk

Makaton
www.makaton.org

Signalong
www.signalong.org.uk

CONTACT A FAMILY

Caring Decisions
From Mary McBride - Regional Development Officer
Contact a Family North West
www.caringmatters.dial.pipex.com

Contact a Family
Regular newsletters and information about a variety of disabilities - support groups etc., for rare disorders.
admin@cafamily.org.uk

Making Contact Org
Contact a Family is due to launch a new website in the summer. Making Contact Org will be dedicated to linking people and families affected by disorders and disabilities on a one-to-one basis.

CONTINENCE

Association For Continence Advice
www.aca.uk.com

DEAF ISSUES

Deafness Support Network
Cheshire, Halton and Warrington.
01606 330633
www.deafnesssupportnetwork.co.uk

National Deaf Children’s Society
www.ndcs.org.uk
Royal National Institute For Deaf People
www.rnid.org.uk

DEPRESSION

Defeat Depression
www.depression.org.uk

Depression Alliance
www.depressionalliance.org

DIRECT PAYMENTS

Database for Personal Assistants
www.Pamatch.co.uk

The Department of Health has published some answers to frequently asked questions about direct payments, including clarification of using a direct payment to employ a relative.
More at www.doh.gov.uk/directpayments/

Instream Partnership, Europa House,
Barcroft Street, Bury BL9 5BT
0161 763 8707
gary@instream.org.uk
john@instream.org.uk
www.instream.org.uk
mobile 07709 490081

West Lancs Peer Support Group
Dresser House
Gillibrands Road
Skelmersdale
WN8-9TU
Tel; 01695-52645
Mobiles 07963-003552 / 0033548

DISABILITY – GENERAL

Abilitynet
www.abilitynet.co.uk

British Council of Disabled People
www.bcodp.org.uk
Council for Disabled Children
www.ncb.org.uk

Disability Alliance
www.disabilityalliance.org

Disability Equality in Education (DEE)
info@diseed.org.uk

Disability Is Natural
www.disabilityisnatural.com

Disability Now
www.disabilitynow.org.uk

Disability Rights Commission
www.drc.gb.org

Disabled Living Foundation
www.dlf.org.uk

Dyspraxia Foundation
www.dyspraxiafoundation.org.uk

European Disability Forum
www.edf-feph.org

The Family Fund
www.familyfund.org.uk

Focus on Disability
www.cross17.freeserve.co.uk

4DP (For Disabled People)
www.4dp.com
info@addiss.co.uk
www.addiss.co.uk

For Parents by Parents
www.forparentsbyparents.com

Handsel Trust
www.handseltrust.org

Hyperactive Childrens Support Group
www.hacsg.org.uk

Joseph Rowntree Foundation
www.jrf.co.uk

Learning Difficulties
www.learningdisabilityuk.org.uk
EDUCATION

C.S.I.E.
Center for Studies on Inclusive Education
www.inclusion.uwe.ac.uk/csie

The Bini materials are a set of four books and video designed to appeal to children who need slower paced video stories with simple language and images. Each video story is 5 minutes long and shares the same beginning and ending with a key event in the middle. A set of four sturdy picture books and a puppet complement the four stories on the video. Each picture book contains the key images on the video.

For more details and prices contact Jennifer Roberts on: 01728 603772 or go on www.meetbini.co.uk

DFES Publication (for free publications on Education issues)
PO Box 5050, Sherwood Park, Annesley, Nottingham NG15 0DJ
Tel: 0845 6022260
fax: 0845 6033360
testphone 0845 6055560
www.dfes@prolog.uk.com
You can also download an electronic version of some reports at:
www.dfes.gov.uk/consultations/

Network 81
1-7 Woodfield Terrace, Stansted, Essex CM24 8AJ
Tel: 01279 647415
www.network81.co.uk

Skill
Chaper House, 18-20 Crucifix Lane, London SE1 3JW
Tel: 0800 3255050
www.skill.org.uk

After 16 – What’s New?
www.after16.org.uk

Listening Books
www.listening-books.org.uk

LEGAL HELP IN EDUCATION

Alliance for Inclusive Education
National campaigning organisation led by disabled people.
ALLFIE: Unit 2, 70 South Lambeth Road London SW8 1RL
Tel:020 7735 5277
Fax: 020 7735 3828
E mail info@allfie.org.uk
www.allfie.org.uk

Bury Parent Partnership
provides independent advisory services to parents regarding the LEA’s process of
identifying assessing and making statements for children with special educational needs.
Bury Parent Partnership, Red Centre, Morley Street, BuryBL9 9JQ.
helpline 0161 763 501
Each authority will have a parent partnership co-ordinator.

Children’s Legal Centre
Runs a free, confidential legal advice service on the rights of the child and their education.
Tel: 01206 873 820

Disability Equality in Education
Training provided by disabled people who are skilled presenters.
Unit GL Leroy House, 436 Essex Road, London NI 3QP
Tel: 020 7359 2855
Fax: 020 7354 3372
email info@diseed.org.uk
www.diseed.org.uk
**Education Lawyers Association**  
Can provide you with the number of a legal professional in your area specialising in education law.  
Tel: 01903 504 949

**IPSEA**  
Independent Panel For Special Educational Advice  
[www.ipsea.org.uk](http://www.ipsea.org.uk)

**Law Centres Federation**  
Can give you the number of your nearest Law Centre.  
Tel: 0170 387 8570

**Parents for Inclusion**  
A national organisation of parents of disabled young people.  
Unit 2 South Lambeth Road, London SW8 1RL  
Tel: 020 7735 7735  
fax: 020 7735 3828  
e mail: info@parentsforinclusion.org  
[www.parentsforinclusion.org](http://www.parentsforinclusion.org)  
Helpline 020 7582 5008

**EMPLOYMENT**

**Association for Supported Employment**  
[www.afse.org.uk](http://www.afse.org.uk)

**First Step Trust**  
[www.fst.org.uk](http://www.fst.org.uk)

**Remploy**  
[www.remploy.co.uk](http://www.remploy.co.uk)

**Shaw Trust**  
[www.shaw-trust.org.uk](http://www.shaw-trust.org.uk)

**EPILEPSY**

**British Epilepsy Association**  
[www.epilepsy.org.uk](http://www.epilepsy.org.uk)  
epilepsy@epilepsy.org.uk

**Epilepsy Action (British Epilepsy Association)**  
[www.epilepsy.org.uk](http://www.epilepsy.org.uk)

**Epilepsy Foundation of America**  
[www.efa.org](http://www.efa.org)
International League Against Epilepsy
www.ilae-epilepsy.org

GOVERNMENT DEPARTMENTS

Commission for Health Improvement
www.chi.nhs.uk

Commission for Racial Equality
www.cre.gov.uk

Criminal Justice System
www.cjsonline.gov.uk

Department for Constitutional Affairs
www.dca.gov.uk

Department of Health
www.doh.gov.uk

Direct Payments
www.doh.gov.uk/directp.htm

Social Exclusion Unit
www.socialexclusionunit.gov.uk

Valuing People
www.valuingpeople.gov.uk

Workforce Development Confederation
www.wdc.nhs.uk

Foetal Alcohol Syndrome
F.A.S.
Foetal Alcohol Syndrome. To find out more contact Margaret Murch.
margiemurch@blueyonder.co.uk

HEALTH ISSUES FOR PEOPLE WITH LEARNING DISABILITIES

Elfrida Society
www.elfrida.com

Health Evidence Bulletins for Learning Disability
www.hebw.uwcm.ac.uk/learningdisabilities

Intellectual Disability Health Information
www.intellectualdisability.info
HEALTH PROMOTION

Health Promise
www.healthpromis.had-online.org.uk

Health Promotion Agency (Northern Ireland)
www.healthpromotionagency.org.uk

My Health My Choice
www.myhealthmychoice.co.uk
www.promotehealth.com

Patient UK Health Promotion
www.patient.co.uk/showdoc.asp?doc=16

UK Wellness
www.ukwellness.com

HUMAN RIGHTS

Human Rights Act 1998

Human Rights Unit
www.humanrightsni.gov.uk

Liberty
www.liberty-human-rights.org.uk

Liberty On Line
www.yourrights.org.uk

Mental Disability Rights International
www.mdri.org

Scottish Human Rights Centre
www.scottishhumanrightscentre.org.uk

VIA (Values Into Action)
Works for the rights of people with learning difficulties.
Tel: 020 7729 5436
e mail general@viauk.org
www.viauk.org/

In Control
In Control is a national programme to change the organisation of social care in England so that people who need support can take more control of their own lives and fulfil their role as citizens. Contact Simon Duffy or Julie Casey.
Valuing People Support Team  
36 Rose Hill Drive  
Moseborough  
Sheffield S20 5PN  
simonduffy@mac.com  
07973 715983  
www.selfdirectedsupport.org

PROMOTING INCLUSION AND CHALLENGING SEGREGATION

Bolton Institute,  
Chadwick Street, Bolton BL2 1JW  
Tel: 01204 903200  
Fax: 01204 903232  
E mail kb@bolton.ac.uk  
www.inclusion-boltondata.org.uk

Inclusion International  
www.inclusion-international.org

Inclusion Network  
www.inclusion.org

Inclusion Press  
www.inclusion.com/index.html

Inclusive Solutions  
www.inclusive-solutions.com

Institute for Community Inclusion  
www.communityinclusion.org

National Resource Centre for Inclusion (USA)  
www.web.syr.edu/%7Ethechp/nrc.htm

LEARNING DISABILITY GENERAL

Association of Practitioners in Learning Disabilities  
www.apld.org.uk

British Institute Of Learning Disabilities  
www.bild.org.uk/

Estia Centre  
www.estiacentre.org

Foundation for People with Learning Disabilities  
www.learningdisabilities.org.uk
Intellectual Disability Network (Australia)
www.monash.edu.au/informatics/idcn.html

International Association for the Scientific Study of Intellectual Disabilities
www.iassid.wisc.edu/index.html

Institute for Health Research
www.lancaster.ac.uk/depts/ihr

Joseph Rowntree Foundation
www.jrf.org.uk

Kings Fund
www.kingsfund.org.uk

Mencap
www.mencap.org.uk

Learning Disabilities UK
www.learningdisabilitiesuk.org.uk

Learning Disability History (Life in institutions)
www.learningdisabilityhistory.com

National Electronic Library for Learning Disability
www.minervation.com/ld

Norah Fry Research Centre
www.bris.ac.uk/Depts/NorahFry/index.htm

North West Training and Development Team
www.nwtdt.com

Paradigm
useful website for all issues
www.paradigm-uk.org

Presidents (USA) Committee for People with Intellectual Disability
www.acf.hhs.gov/programs/pcpid/index.html

Royal Society of Medicine Learning Disability Forum
www.rsm.ac.uk/academ/forld.htm

Scottish Consortium for Learning Disabilities
www.scld.org.uk

Standing Conference of Voluntary Organisations for People with a Learning Disability in Wales
www.scovo.org.uk

Tizard Centre
www.ukc.ac.uk/tizard/
UK Learning Disabilities
www.uklearningdisabilities.co.uk

Vanderbilt Kennedy Centre for Research on Human Development
www.kc.vanderbilt.edu/kennedy

MENTAL HEALTH ISSUES

Mental Health Care
www.mentalhealthcare.org

Mental Health Europe
www.mhe-sme.org

Mental Health Foundation
www.mentalhealth.org.uk/

Mental Health Internet Resources
www.mentalhealth.com

Mental Health Matters
www.emental-health.com

Mental Health Media
www.mhmedia.com

Mental Health Professional Resources
www.mentalhelp.net/prof.htm

Mental Health Sanctuary
www.mhsanctuary.com

Mental Health Web Network
www.mentalhelp.net/

Mental Help Net
www.mentalhelp.net

Mind
www.mind.org.uk/

Mind Out for Mental Health
www.mindout.net

National Electronic Library for Mental Health
www.nelmh.org

National Institute for Mental Health England (NIMHE)
www.nimhe.org.uk

On Line Dictionary of Mental Health
www.shef.ac.uk/~psych/psychotherapy/index.html
Primary Care Mental Health and Education
www.primhe.org

Sainsbury’s Centre for Mental Health
www.scmh.org.uk

SANE
www.sane.org.uk

Scottish Development Centre for Mental Health
www.sdcmh.org.uk

University of Adelaide Mental Health and Psychiatry Internet Resources
www.library.adelaide.edu.au/guide/med/mentalhealth

Wales Centre for Mental Health Service Development
www.walescfmhsd.org.uk

World Association for Psychosocial Rehabilitation
www.wapr.net/frame.html

MENTAL HEALTH ACT

Institute of Mental Health Law
www.imhl.com/

Institute of Mental Health Act Practitioners
www.markwalton.net

Mental Health Act Commission
www.mhac.trent.nhs.uk

Mental Health Act Guide
www.hyperguide.co.uk/mha/

MENTAL HEALTH IN LEARNING DISABILITY

California Mental Health and Developmental Disabilities Centre
www.npi.ucla.ed/mhdd

Estia Centre
www估stiacentre.org

Institute for Dual Diagnosis
www.theraed.com

National Association for the Dually Diagnosed (NADD)
www.thenadd.org
MENTAL HEALTH PROMOTION

History of Learning Disability Nursing
www.shef.ac.uk/~nmhuk/ldnurs/ldnhome.html

International Network for Mental Health Promotion
www.mhpconnect.com/index_e.html

National Network for Learning Disability Nurses
www.nnldn.org.uk

OCCUPATIONAL THERAPY

British Association of Occupational Therapists
www.cot.co.uk

OFFENDING BEHAVIOUR

Forensic Nursing Resource
www.fnrh.freeservce.co.uk

International Association of Forensic Mental Health Services
www.iamhs.org

Revolving Doors
www.revolving-doors.co.uk

What Works with Offenders?
www.whatworkswithoffenders.co.uk

PERSON CENTRED PLANNING

Circles Around Dundee
www.circlesarounddundee.org.uk/pcp

NWDTDT (North West Training and Development Team)
www.nwtdt.com

Person Centred Planning Educational Site
www.ilr.cornell.edu/ped/tsal/Enable
PSYCHIATRY

Royal College Of Psychiatry
www.rcpsych.ac.uk/index.htm

World Psychiatric Association
www.wpanet.org

PSYCHOLOGY

Academy of Cognitive Therapy
www.academyofct.org

Association for Psychological Therapists
www.apt.ac/

Beck Institute for Cognitive Therapy
www.beckinstitute.org

British Association for Behavioural and Cognitive Psychotherapies
www.babcp.org.uk

British Association for Counselling and Psychotherapy
www.counselling.co.uk

British Psychoanalytical Society
www.psychoanalysis.org.uk

British Psychological Society
www.bps.org.uk/

Cognitive Behaviour Therapy
www.cognitivetherapy.com

Psychology Network
www.psychnet-uk.com/

UK Council for Psychotherapy
www.psychotherapy.org.uk

RESIDENTIAL SERVICE PROVIDERS

Care Principles
www.careprinciples.com

Choice Support
www.choicesupport.co.uk
Home Farm Trust  
www.hft.org.uk

Housing Options  
www.housingoptions.org.uk

Hyde Housing Association  
www.hyde-housing.co.uk

Larche  
www.larche.org.uk

Ling Trust  
www.lingtrust.org.uk

Partnerships in Care  
www.partnershipsincare.co.uk

United Response  
www.unitedresponse.org.uk

SELF HARM/SELF INJURIOUS BEHAVIOUR

Secret Shame  
www.palace.net/~llama/psych/injury/html

SENSORY IMPAIRMENTS
Tel: 01606 330633  
www.deafnesssupportnetwork.co.uk

National Deaf Services  
www.nationaldeafservices.com

Royal National Institute for Deaf People  
www.rnid.org.uk

Royal National Institute of the Blind  
www.rnib.org

SENSE  
www.sense.org.uk

SERIOUS MENTAL ILLNESS

Hearing Voices Network  
www.hearing-voices.org

Rethink  
www.rethink.org
Schizophrenia
www.schizophrenia.com

World Fellowship for Schizophrenia and Allied Disorders
www.world-schizophrenia.org

SPEECH AND LANGUAGE THERAPY

Royal College of Speech and Language Therapists
www.rcslt.org

SOCIAL WORK

British Association of Social Workers
www.basw.co.uk

SYNDROMES

Angelman’s Syndrome
www.asclepius.com/angel

Birth Defects Foundation
www.birthdefects.co.uk

Cornelia de Lange Syndrome
www.cdls.org.uk

Down’s Syndrome Association (UK)
www.dsa-uk.com

Down’s Syndrome Association (USA)
http://nas.com/downsyn/index.html

Dysmorphic Syndromes
www.hgmp.mrc.ac.uk/DHMHD/view_human.html

Fragile X Research Foundation
www.fraxa.org

Fragile X Society
www.fragilex.org.uk

Genes & Disease

Klinefelter’s Syndrome
www.ksa-uk.co.uk
Prada Willi Association (UK)
www.pwsa-uk.demon.uk

Prada Willi Association USA
www.pwsausa.org

Rare Genetic Diseases in Children
www.med.nyu.edu/rgdc/homenew.htm

Rett’s Syndrome Association (UK)
www.rettsyndrome.org.uk

Smith-Magenis Syndrome
www.kumc.edu/gec/support/smith-ma.html

Tourette’s Syndrome
www.tsa.org.uk

Tuberous Sclerosis Association
www.tuberous-sclerosis.org

Turner’s Syndrome UK
www.tss.org.uk

Turner’s Syndrome (USA)
www.turner-syndrome-us.org

UK Resources for Down’s syndrome
www.43green.freeserve.co.uk/uk_downs_syndrome/ukdsinfo.html

Williams Syndrome
www.williams-syndrome.org.uk

HOUSING

National Centre for Independent Living (NCIL)
Information and advice on independent living, direct payments.
Tel: 020 7587 7587 1663
Text. 0207587 1177
e mail ncil@ncil.org.uk
www.ncil.org.uk

Norah Fry Research Centre
Living it up Free Magazine and CD
3 Priory Road, Bristol BS8 1TX
Tel: 0117 923 8137
minicom 0117 928
Fax 0117 946 6553
Valuing People Support Team
C/O NWTDT office
The Globe Centre
St James Square
Accrington  BB5 0RE  Tel 254 306858
www.housingoptions.org.uk

National forum site, the site for the white paper valuing people
www.valuingpeople.org.uk

Government site for carers
www.carer.gov.uk

A source of information on Social Role Valorisation the values base for person centred planning.
www.communityliving.org.uk

www.soeweb.syr.edu/thechp-/rsapub

One of the leading providers of supported living in the USA.
www.optionsmakison.com

For information and resources in all issues concerning inclusion.
www.inclusion.com

Probably the best site in learning disability services-lots of good links.
www.paradigm-uk.org

Website for Essential Lifestyle Planning.
www.allenshea.com/familyplan

Citizen Advocacy information and training site.
www.citizenadvocacy.org.uk

Website for the helpline promised in valuing people.
www.mencapp.org.uk/helpline

Scotland wide leading organisation, -runs partners and other leadership courses.
www.shstrust.org.uk

A grassroots advocacy organisation in Virginia USA-good articles.
www.commcoal.org

A great site for inspiring articles- see especially ‘toolbox for change’.
www.dimagine.com

The website for community service volunteers
www.csv.org.uk

A great website for the north west, downloads on essential lifestyle planning and lots more. Many leadership development courses.
www.nwttdt.com
Website for a local person centred planning facilitators group
www.speargroup.org.uk
For up to date information on direct payments in Scotland.
www.dpscotland.org.uk

Website for Norfolk people first group.
www.peoplefirstnorfolk.org

Website on research and issues surrounding parents with learning disabilities.
www.supported-parenting.com

Supported Employment Association
www.afse.org.uk

A housing advisory service for people with learning disabilities.
www.housingoptions.org.uk

The quality network helps people to check the quality in services for people with learning disabilities.
www.bild.org.uk/quality_network2Qnhomepage.htm

Keeping It Real
Keeping It Real is an organisation formed to explore how the use of ICT and other innovative methods can be used when planning with people with additional needs.
www.KeepingItReal.org

There is the Transition Champions toolkit on VPST web site. To help learning disability partnership boards improve their support for family carers. Entitled valuing families: a toolkit for family friendly services.

H.F.T. (Home Farm Trust)
Have developed a pack on ‘Transition’.
emma.nichols@hft.org.uk

The Social Services Inspectorate:
A history - Origins, impact and legacy is a report commissioned to mark the 19 years that the Social Services Inspectorate has been part of the Department of Health. It can be downloaded from:
http://www.dh.gov.uk/assetRoot/04/07/75/95/04077595.pdf

Sure Start
Caxton House, 6-12 Tathill Street, London SW1H 9NA
Tel: 0870 0002288
Fax: 020 7273 5124
www.surestart.gov.uk
info.surestart@dfes.gsi.gov.uk
STROKES
Different Strokes
Support for stroke survivors. Different Strokes has contacts in most areas of the UK, they produce regular newsletters, very positive outlook.
9 Canon Court, Wolverton Mill, Milton Keynes MK12 5NF
Tel: 0845 130 7172
Fax: 01908 313 501
www.differentstrokes.co.uk
E-Mail info@differentstrokes.co.uk