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
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Contents

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

DIAGNOSIS / CLASSIFICATION

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

Syndrome	Audiovisual	Endocrine	Psychiatric / Psychological	CNS
Cerebral Palsy **<1:500**	Visual Impairment Hearing Impairment	-	Depression Variable intellectual capacity	
	Cardiovascular	Muscular/ Skeletal	Other	Inheritance
		Orthopaedic problems Neuromuscular problems	Genito-urinary problems Incontinence Constipation Dental problems Recurrent aspiration Oesophagitis Gastroesophageal Reflux +/- bleeding/ anemia Swallowing / eating diffics.	

Syndrome	Audiovisual	Endocrine	Psychiatric / Psychological	CNS
Down Syndrome **<1:700**	Visual impairment (multifactorial) cataracts Hearing impairment (multifactorial) (Annual assessments recommended)	Hypothyroidism (Annual TFT recommended)	Depression Alzheimer's type dementia (Clinical onset uncommon before 40 years)	
	Cardiovascular	Muscular/ Skeletal	Other	Inheritance
	Congenital Heart Defects (common in 40 to 50%)	Atlantoaxial instability Skin disorders, alopecia, eczema	Blood dyscrasias Childhood leukaemia Sleep apnoea Increased susceptibility to infections Coeliac disease	Most cases are sporadic; 4% due to translocation involving chromosome 21 or rarely parental mosaicism

Syndrome	Audiovisual	Endocrine	Psychiatric / Psychological	CNS
Prader-Willi	Strabismus Myopia	NDDM (secondary to obesity) Hypogonadism Delayed puberty	Hyperphagia Impulse control difficulties Self-injury	
<1:10,000-25,000	Cardiovascular	Muscular/ Skeletal	Other	Inheritance
		Scollosis, Kyphosis Hypotonia Skin picking	infantile failure to thrive, then hyperphagia and severe obesity High tolerance to pain Decreased ability to vomit Sleep apnoea Osteoporosis Undescended testes Dental Abnormalities	Atypical. Most cases are sporadic

Syndrome	Audiovisual	Endocrine	Psychiatric / Psychological	CNS
Fragile X	Visual impairment Hearing impairment Recurrent ear infections		Attention deficit/hyperactivity Variable intellectual capacity Disabled in social functioning	
<1:6,000	Cardiovascular	Muscular/ Skeletal	Other	Inheritance
	Aortic dilation, Mitral Valve prolapse (related to connective tissue dysplasis	Connective tissue dysplasia Scoliosis Congenital Hip Dislocation	Herniae (CT related) Abnormalities of speech and language	X linked

Syndrome	Audiovisual	Endocrine	Psychiatric / Psychological	CNS
Angelmann Syndrome	Glaucoma		Easily excitable Hyperactive	Severe developmental delay Epilepsy
<1:10,000	Cardiovascular	Muscular/ Skeletal	Other	Inheritance
		Joint contractures and scoliosis (in adults)	Speech impairment Movement and balance disorder Characteristic EEG changes	Variety of genetic mech-anisms on Chromosome 15

Syndrome	Audiovisual	Endocrine	Psychiatric / Psychological	CNS
Williams	Hyperacusis Strabismus		Variable intellectual capacity Attention deficit problems in childhood	Perceptual and motor function reduced
?<1:20,000	Cardiovascular	Muscular/ Skeletal	Other	Inheritance
	Cardia abnormalities Hypertension, CVAs Chronic hemiparesis	Joint contractures Scoliosis Hypotonia	Renal abnormalities	Microdeletion on chromosome 7

Syndrome	Audiovisual	Endocrine	Psychiatric / Psychological	CNS

Syndrome	Audiovisual	Endocrine	Psychiatric / Psychological	CNS
Rett	Refractory erros		Severe intellectual disability	Epilepsy Vasomotor instability
<1:14,000 Female	Cardiovascular	Muscular/ Skeletal	Other	Inheritance
	Prolonged QT Interval	Osteopenia Fractures Scoliosis	Hyperventilation Apnoea Reflux Feeding difficulties Growth failure	Usually sporadic X linked

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Syndrome	Audiovisual	Endocrine	Psychiatric / Psychological	CNS
Noonan	Strabismus, refractive errors Vision/hearing Impairments		Mild intellectual disability	Epilepsy
	Cardiovascular	Muscular/ Skeletal	Other	Inheritance
<1:10,000	Pulmonary Valvular Stenosis ASD, VSD, PDA	Scoliosis Talipes equinovarus Pectus carinatum/excavtum	Abnormal clotting factors, platelet dysfunction Undescended testes, deficient spermatogenesis Lyphoedenoma Hypatosplenomegaly Cubitus valgus, hand abnormalities	Autosomal dominant may be sporadic

Syndrome	Audiovisual	Endocrine	Psychiatric / Psychological	CNS
Tuberous Sclerosis	Retinal tumours Eye rhabdomyomates		Variable intellectual capacity Behavioural difficulties Sleep problems	Cerebral astrocytomas Epilepsy
	Cardiovascular	Muscular/ Skeletal	Other	Inheritance
<1:6,000 – 17,000	Rhabdomyomates Hypertension	Bone Rhabdomyomata	Kidney and lung harmartomata Polycystic kidneys Liver Rhabdomyomata Dental abnormalities Skin lesions	Autosomal dominant

Adapted from an original unpublished version by Michael Kew and Glyn Jones – The University of Queensland
Page last updated on

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 (Wideset eyes with droopy eyelids)
Pulmonary stenosis
Ventricular septal defect
Bracydactly 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

Acrodysostosis

ALTERNATIVE 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:

Short stature
Distinctive craniofacial features:
Small head circumference
Telecanthus or ocular hypertelorism
Small nose
Tented upper lip
Everted lower lip
Urogenital anomalies ranging from hypospadias and undescended testicles to severe hypospadias and ambiguous genitalia - prone to urinary tract infections
Cardiovascular abnormalities in some cases
Epilepsy
Skeletal abnormalities can be quite diverse
Uncoordinated swallow
Constipaton and poor bladder and bowel control
mild form of hemoglobin H disease

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Mild to severe learning disabilities
Absent speech

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 famial 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

ALTERNATIVE NAMES:

Neuronal ceroid lipofuscinosis
Juvenile Batten Disease

GENERAL HEALTH SCREENING:

Ophthalmic assessment
Monitoring epilepsy and treatment
Monitoring of mental health

PHYSICAL PHENOTYPE:

Ataxia (Gradual onset)
Seizures (myoclonic/tonic)
Retinal degeneration

Associated Complications:
Spasticity
Visual impairment
Kyphosioliosis

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Learning Disabilities (Gradual Intellectual Decline)

Associated Complication:
Psychosis

CONTACT:

Batten Disease Family Association
www.bdfauk.freeserve.co.uk

www.RNIB.org.uk – Visual impairments

Batten support and Research Trust
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 Conneticut Avenue
NW, Suite 404
Washington DC
20008-234
USA

Tel: 2029665557
www.geneticalliance.org

Branchio –Oculo-Facial Syndrome

ALTERNATIVE NAMES:

BOFS
Branchiooculartacial 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/pollices
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 agenesis
Scalp tumours
Short palpebral fissures,short philtrum
Stature/length short proportunate
Skin atrophy - patchy
Small/yhpoplastic/deepset nails/claws
Telangiectasia/angiokeratomata of skin

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Mild learning disability in some cases

CONTACT:

WWW.ABILITY.ORG.UK

Cornelia de lange syndrome

ALTERNATIVE NAMES:

De- Lange Syndrome
Cornelia de Lange Syndrome
Amsterdam Dwarfism

GENERAL HEALTH SCREENING:

Hearing Assessment
Cardiovascular Assessment
Ophthalmic assessment
Monitoring of Respiratory function
Monitoring and treatment of respiratory infections
Monitoring of Male Genitalia
Hearing assessment
Gastroeosophogeal assessment

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 Syndactly&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

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 visula fields, acuity.
Sensitivity to light
40% facial palsy may be accompanied by sensorineural hearing loss
30% swallowing problems
Heart defects - verying 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

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 mal formed ears
Hypertelorism
Epicanthal folds prominent philtrum
Broad nasal root
High arched palate-possible cleft lip or palate
Flattened chest with widely spaced nipples
Omphalocele
Clindactly
Brachydactyly
Camptodactly and tapered fingers
Dermatoglyphic defects
Hypotonia
Hypertrichosis and hypoplastic nails
Epilepsy
Crainosynostosis
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, straght nasofrontal angle, short philtrum and epicanthal folds
Umbilical or inguinal hernia
Rockerbotton feet, thumb deformities, syndactyly, abnormal dermatoglypghics
Scoliosis
Hypertonia or hypotonia
Epilepsy
Heart murmour
Venous return anomalies
Horseshoe kidney
Renal hypoplasia
Urethro-vesical reflux
Criptorchidism
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: Maxillaryhypoplasia
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
Hypothrnar 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
Occcasionaly 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:

GENERAL HEALTH SCREENING:

Dietetic assessment
Ophthalmic assessment
Assessment and monitoring of epilepsy
Maxillofacial assessment

PHYSICAL PHENOTYPE:

Microcephaly
Short philtrum
Prominent upper central incisors
Palpebrl fissures slant down
Narrow and high arched palate
Microganthia
Maxillary hypoplasia
Truncal obesity
Short stature
Arachnodactly
Narrow hands and feet
Joint laxity muscle weakness/myopathy
Ophthalmic irregularities, myopia strabismus
Pigmentatary retinopathy/chroioretinitis
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
sesitivity 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 syndactly
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

5p minus society – family support group
Web: 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 shapedhead), brachycephaly, palable ridging, flat occiput, high prominent forehead, flattened face with maxillary hypoplasia, relative mandibular prognathism,low set ears - conductive hearing loss, downslanting 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 preauriccular 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
Clinodactly/polydactly
Rocker bottom feet
Abnormal insertion of thumbs
Hypotonia
Heart murmur
Crytorchidism
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

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:

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

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Learning disability

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:srabismus, 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 carinoma
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

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 thrombocytopnia

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 oppicut, brachycephaly and small maxilla
Abnormal auricles and absent lobules
Upslanting eyes
Palpebral fissures/brushfield spots
Strbismus 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

DETAILED OUTLINE OF HEALTH NEEDS FOR PEOPLE WHO HAVE DOWNS SYNDROME

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:

3. Catalano RA (1990) *Down syndrome* Survey of Ophthalmology, 34, 385-398.
7. Traboulsi EI, Levine E, Mets MB, Parelhof ES, O'Neill JF, Gaasterland DE (1988) *Infantile glaucoma in Down's syndrome(trisomy 21)* American Journal of Ophthalmology,105, 389-394.
8. Cullen J, Butler H (1963) *Mongolism(Down's syndrome) and keratoconus* B.J. Ophthalmology. 47. 321-330
9. Volker-Dieben,HJ.,Odenthal,MTP.,D'Amaro,J.,KruitPJ.(1993) *Surgical treatment of corneal pathology in patients with Down's syndrome* J.Int.Disability.Res. 37 167-175

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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 3.
- Hearing impairment can be successfully managed in this population. If undetected it is likely to be a significant cause of preventable secondary handicap (3.21).
- 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.(3)
- Because of an increased incidence of congenital sensorineural loss newborns should be included in neonatal screening programmes (14).
- At all ages people with Down's syndrome have narrow ear canals which predispose to accumulation of wax 4. 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 (22).
- Diagnostic Auditory Brain Stem (ABR) responses in people with Down's syndrome must be interpreted with caution (22)
- In adults with the syndrome hearing assessment is essential in the differential diagnosis of depression and dementia (7).

References

3. Cunningham,C.,McArthur,K.,(1981) Hearing loss and treatment in young Down's syndrome children. Child: care,health and development. **7** : 357-374.
4. Dahle,AJ.,McCollister,FP.,(1986) Hearing and otologic disorders in children with Down syndrome. American Journal of Mental Deficiency. **90** (6) : 636-642.
5. Davies,B.,Penniceard,RM.(1980) Auditory function and receptive vocabulary in Down's syndrome children. In.'Disorders of Auditory Function lll' Eds Taylor,IG.,Markides,A. Academic Press.
7. Evenhuis,HM.,van Zanten,GA.,Brocnar,MP.,Roerdinkholder,WHM.(1992) Hearing loss in middleage persons with Down syndrome. Am.J.on Mental Retardation.**97**.47-56.

14. National Deaf Children's Society (1994) Quality standards in Paediatric audiology. Volume 1. Guidelines for early identification of hearing impairment. ISBN 0904691 36 5
21. Whiteman,BC.,Simpson,GB.,Compton,WC.(1986) Relationship of otitis media and language impairment of adolescents with Down's syndrome. Mental Retardation.**24.** (6) : 353-356.
22. Widen,JE.,Folsom,RC.,Thompson,G.,Wilson,WR.(1987) Auditory brainstem responses in young adults with Down syndrome. Am.J.Mental Deficiency.**91.**472-479

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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 atrioventricular septal defects.(1.2) 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.(3)
- 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 (2.8.11)
- 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 (9.14).
- 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:

1. Tubman.,TRJ.,Shields,MD.,Craig,BQ.,Mulholland,HC.,Nevin,NC., (1991) *Congenital heart disease in Down's syndrome; Two year prospective early screening study*. BMJ; 302 : 1425-1427.
2. Frid,C.,Drott,P.,Lundell,B.,Rasmussen,F.,Anneren,G. (1999). *Mortality in Down's syndrome in relation to congenital malformations.* J.Int.Disab.Res. 43.3.234-241
3. Amark,K.,Sunnegarth,J (1999) *The effect of changing attitudes to Down's syndrome in the management of complete atrioventricular septal defects.* Arch.Dis.Ch.81.2. 151-154
8. Taylor,JFN.,(1990) *Commentary : Natural and modified history of atrioventricular canal defect - a 17 year study*. Arch Dis. Child., 65: 966-967.
9. Committee Report, 1995. *Guidelines for optimal medical care of persons with Down syndrome*. Acta Paediatrica, 84 : 823-827.
11. Wren,C.,Richmond,S.,Donaldson,L. (1999) *Presentation of congenital heart disease in infancy: implications for routine examination.* Arch.Dis.Child.Fetal Neonatal Ed. 80 F49-
14. Goldhaber,SZ.,Brown,WD.,St. John Sutton,MG., (1987). *High frequency of mitral valve prolapse and aortic regurgitation among asymptomatic adults with Down syndrome*. JAMA : 258; 13; 1793-1795

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 hydrothyroidism 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:

1. Cutler A T, Benezra-Obeiter R, Brink S J, (1986). *Thyroid function in young children with Down's Syndrome.* Am J Dis Child. 140. 479-483

2. Pueschel S M, Jackson I M D, Giesswein P, et al. (1991). *Thyroid function in Down's Syndrome.* Res Dev Disabil Res. 12. 287-296
3. Takahashi H, Bordy M D, Sharma V, Grunt J A, (1979). *Hyperthyroidism in patients with Down's syndrome.* Clinical paediatrics; 18. 273-275.

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

1. Davidson,RG.,(1988). *Atlantoaxial Instability in Individuals With Down Syndrome: A Fresh Look at the Evidence.* Pediatrics: 81: 857-865
2. Saad,KFG., (1995) *A lethal case of atlantoaxial dislocation in a 56-year-old woman with Down's syndrome.* J.Intellectual. Disability Research. 39: 447-449
4. Cremers,MJG.,Ramos,L.,Bol,E.,van.Gijn,J. (1993) *Radiological assessment of the atlantoaxial distance in Down's syndrome.* Archives of Disease in Childhood. 69.347-350.
6. Department of Health.(1995). *Cervical spine instability in people with Down syndrome.* CMO Update 7.p4.
9. Casey,AT.,O'Brien,M.,Kumar,V.,Hayward,RD.,Crockard,HA. (1995) *Don't twist my child's head off : iatrogenic cervical dislocation*. BMJ. 311: 4 November. p1212-1213.

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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:

1. McCoy EE.(1992) *Growth Patterns in Down's Syndrome*. In Down Syndrome: Advances in Medical Care, Ed. Lott IT, McCoy EE. Wiley-Liss, Inc. NewYork ISBN 0471561843.
3. Greenwood RD, Nadas AS (1976); *The clinical course of cardiac disease in Down's syndrome.* Pediatrics 58: 893:897.
4. Stebbens VA, Samuels MP, Southall DP, Dennis J, Croft CB; (1991)*. Sleep related upper airway obstruction in a cohort with Down's syndrome*. Arch.Dis.Child. **66.**1333-1338
5. George,EK.,Mearin,ML.,Bouquet,J.,von Blomberg,BME., et al (1996) *High frequency of coeliac disease in Down syndrome* J.Pediatr. **128.** 555-557
7. Karlsson B, Gustafsson J,Hedov G, Ivarsson S-A,Anneren G (1998) *Thyroid dysfunction in Down's syndrome:relation to age and thyroid autoimmunity*. Arch. Dis Childhood.79.242-245.
9. *Growth charts for children with Down's syndrome*. Harlow Printing. South Shields NE33 4PU. UK
10. Styles ME, Cole TJ, Dennis J, Preece MA (2001) *New cross sectional stature, weight and head circumference references for Down's syndrome in the UK and Republic of Ireland.* Arch.Dis.Childhood. In Press.

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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 decongester
    - 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:
    - Disordered bowel function tending to diarrhoea or to new onset constipation
    - Failure to thrive as indicated using Down's syndrome specific reference charts (Harlow Printing 2000);
    - Abdominal distension
    - General unhappiness and misery
    - Arthritis
    - Rash suggesting dermatitis herpetiformis
    - 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

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:

Strafstrom, C.E., Patxot, O.F., et al. (1991). Seizures in children with Down syndrome; etiology, characteristics, and outcome. Developmental Medicine and Child Neurology, 33, 191-200.

Stafstrom et al. (1993). Epilepsy in Down syndrome: clinical aspects and possible mechanisms. American Journal of Mental Retardation, supplement, 12-26.

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:

Edwards, J.P. (1990). Sexuality, marriage, and parenting for persons with Down syndrome. In: S.M. Pueschel (Ed.) *The young person with Down syndrome*. Baltimore: Paul Brookes. 187-204

Elkins, T.E., McNeeley, D.G., Rosen, D., et al. (1988). A clinical observation of a program to accomplish pelvic exams in difficult-to-manage patients with mental retardation. *Adolescent Pediatr Gynecol*, 1, 195-198.

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:

Nespoli et al. (1993). Immunological features of Down's syndrome: a review. *Journal of Intellectual Disability Research*, 37, 543-551.

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:

Prasher,V.,Smith,B (2002) Down Syndrome and Health Care. BILD publications.

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:

Cooper, S.A., & Collacott, R.A. (1994). Clinical features and diagnostic criteria of depression in Down's syndrome. *British Journal of Psychiatry*, 165(3), 399-403.

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
Ophthalmic assessment
Dermatological assessment
Assessment of hearing
Dental 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
Brachucephaly
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:

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

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

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
Micropthalmia
Blepharophimosis
Sclerocornea coloboma
Short nose with aflat bridge
Cleft lip or palate
Camptodactyly, syndactly, clinodactly
Scoliosis/kayphosis
Hypotonia
Heart defects
Cryptorchidism
Kidney aplasia or hypoplasia

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

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
Strabims
Cataracts
Microphthalmia
Prominent bulbous nose
Cleft lip and palate
Highly arched palate
Inguinal and diaphragmatic hernia
Camptodactly
Rocker bottom feet
Joint and laxity contractures
Scoliosis and kyphosis
Café-au-lait patches
Congenital heart defects
Imperforate anus/jejunal artesia
Crpotocrhidism 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:	**PSYCHOLOGICAL/BEAHVIOURAL PHENOTYPE:**
Short stature Bulging of the chest sternum Flattening of the vertebrae and upper border of the pelvis (Iliac crest) Shortening of the metacarpals	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 unrinary 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 and hypoplasia
Hypotonia
Bone defects similar to those in chondrodysplasia punctata
Puntata epiphyses
Calcifcation disorders with stippling if the epiphyses
Brain agesis
Hydrocephalus
Agesis of the corpus callosum
Meningoencephalocele 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

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: bracydactly and clinodactly - 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

Foetal alcohol syndrome

ALTERNATIVE NAMES:

GENERAL HEALTH SCREENING:

Cardiac Monitoring
Hearing assessment
Ophthalmic assessment

PHYSICAL PHENOTYPE:

Growth retardation: head, weight and height

Mircocephaly
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 re atrial septal defect
Limb abnormalities of joints, hands, feet, fingers and toes.
Recurrent infections, mostly connected with the respiratory tract

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Learning Disability
Hyperactivity

Contact:

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

Tel: 0151 284 2900

www.medicouncilalcol.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
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
Aanophthalmia

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Learning Disability

Contact:

FACES: The National Cranio Facial Association
USA
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

Goldenhars 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

PHYSICAL PHENOTYPE:

Hydrocephalus
Thickening of the nostrils
Short neck
Short stature
Mild kyphosis
Upper airway obstruction
Recurrent rhinorrhoea obstructive airway disease
Pulmonary hypertention
Cardiac functioning: valvular dysfunction, myocardial thickening, coronary artery narrowing, myocardial infarction
Chronic diarrhoea
Inguinal/umbilical hernias
Severe retinal deterioration & pigmentation
Recurrent otis media -progressive hearing loss
Nodules of growth over the skin of the shoulders, scapulas, posterior chest wall and arm
Epilepsy

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Learning Disability
Hyperactivity
Aggressive Behaviour
Mental Deterioration

Contact:

International Rare Disease Support Network
www.raredisorders.com

or information at,
www.nlm.nih.gov/medlineplus/ency/articale/001203.htm

Hurlers Syndrome

(Life expectancy approx. 10 years)

ALTERNATIVE NAMES:

Mucopolysaccharidosis typel
Alpha-L-Iduronase 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 Nucopolysaccharide Diseases
46 Woodside Road#Amersham
Buckinghamshire
HP6 6AJ

Tel: 01494 434156

Email: mps@mpssociety.co.uk
Web: www.mpssociety.co.uk
www.fabry.org.uk

Johanson Blizzard Syndrome

ALTERNATIVE NAMES:

JBS
Nasal Alar Hypoplasia, HypoThyroidism, Pancreatic Achylia, Cong. Deafness
Ectodermal Dysplasia -Exocrine Pancreatic Insufficiency
Malabsorption-Ectodrmal 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/ dextrcardia, 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
Learge 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
Polydactly
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

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
Arachnodactly, camptodactly,talipes equivarus 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 Blephar optosis
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

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
Laryngotracheal 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

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 phenylananine's role as precuror of melanin)
Inability to process the amino acid phenylalanine
Microcephaly (MPKU also)
Congenital heart defects (MPKU also)
Low birth weight (MPKU only)
Poor co0ordination
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
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 dentalcaries/enamel hypoplasia
Hypogonadism/cryptorchidism
Insatiable appetite
Reduced calorie intake requriement
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

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:

<u>Occurs mainly in females although can on rare occasions occur in males</u>
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 iiii (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/

Or

Contact a Family
Tottenham Court Road
London
W1P 0HA

Tel: 020 73833555
Web: 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
Epiliepsy - 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 Sydnrome
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
Polydactly
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
Microganthia
Pyloric stenosis
Hirshprung 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 injurous 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:

Facial changes are evident with age from rounded in infancy to long and thin with age
Hypotonia may persist
Joint laxity and pes planus
Speech impairment
Macrocrania
Disproportunately large head with a slightly protrusive forehead
Large hands and feet
High arched palate
Premature eruption of teeth
Sparseness of hair
Hypertelorism
Downslanting eyes
Clumsiness, awkward gait
Constipation
Persistent drooling and reluctance to chew
Puberty: Females - early menarche. Males delayed puberty
Frequent respiratory infections - often resulting in conductive hearing loss
Cerebral: ventricular dilation
Seizures - possibly only febrile
Wilms Tumour – renal tumour

PSYCHOLOGICAL/BEHAVIOURAL PHENOTYPE:

Mild to severe Learning Disability
Aggressive behaviour
Ritualistic behaviour
Sleeping problems
Attention deficits
Hyperactivity

Contacts:

Soto's Group Child growth Foundation
2 Mayfield Road
Chiswick
W4 1PW

Tel: 0181 9950257

Or

Soto's Syndrome Support Association

Email: sssa@well.com
Web: www.well.com/user/sssa/

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
Opthalmologic assessment
Echocardiography screening
Electrocardiograms (if abnomal heart rhythm)
CTScan for lesions on lung
Neurodevelopment testing

PHYSICAL PHENOTYPE:

Epilepsy
Skin discolouration, 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
Tuberous Sclerosis Association – www.tuberous-sclerosis.org
Contact a family – 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
Camptodactly
Clinodactly
Short metacarpal/tarsals other anomalies
Absent or dysplasicpatellae
Multiple joint contractures
Abnromal 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

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

Muscloskeletal 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
Email: 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 AROUNG 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.aspergersyndrome.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.deafnessupportnetwork.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

Learning Disabilities
www.learningdisability.co.uk

Learning Disabilities Links
www.rnld.co.uk

Mental Health Foundation
www.mentalhealth.org.uk

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

Partners In Care
www.partnersincare.co.uk

People In Action
www.people-in-action.co.uk

Radar: The Disability Network
www.radar.org.uk

Rett Syndrome Association UK
www.rettsyndrome.org.uk

The Scoliosis Association UK
www.sauk.org.uk

UniquelyGifted
www.uniquelygifted.org

Whizz Kidz
www.whizz-kidz.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 beginnig and ending with a key event in the middle. A set of foursturdy picture books and a puppet complenment 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, confidetial 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 N! 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 yhour area specialising in education law.
Tel: 01903 504 949

IPSEA
Independent Panel For Special Educational Advice
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
Helpline020 7582 5008

EMPLOYMENT

Association for Supported Employment
www.afse.org.uk

First Step Trust
www.fst.org.uk

Remploy
www.remploy.co.uk

Shaw Trust
www.shaw-trust.org.uk

EPILEPSY

British Epilepsy Association
www.epilepsy.org.uk
epilepsy@epilepsy.org.uk

Epilepsy Action (British Epilepsy Association)
www.epilepsy.org.uk

Epilepsy Foundation of America
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
www.hmso.gov.uk/acts/acts1998/19980042.htm

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.ed/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.estiacentre.org

Institute for Dual Diagnosis
www.theraed.com

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

PAS-ADD
www.web.onetel.com/~drplee

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

NWTDT (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
Deafness Support Network. Cheshire ,Halton and Warrington.
Tel: 01606 330633
www.deafnessupportnetwork.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
www.ncbi.nlm.nih.gov/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

A source for articles by John O' Brien and Connie Lyle O'Brien.
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.
wwww.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.nwtdt.com

Website for a local person centred planning facilitators group
www.speargroup.org.uk
For up to date information on direct payments inScotland.
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
Free early education advice. Leaflets and information. Health and family support.
Department for Education & Skills and Department for Work and Pensions
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