

ORAL MANIFESTATIONS IN GENETIC SYNDROMES WITH MENTAL RETARDATION

Purpose

The purpose of this module is to acquaint dental professionals with the characteristics of the major syndromes encountered on an institutional setting with particular emphasis on dental implications.

Learning Objectives

After reviewing this material, the participant should be able to:

1. List 5 classifications of genetic syndromes.
2. List 2 specific syndromes within each category and their significant oral manifestations.
3. Describe the facial characteristics associated with the illustrated syndromes.

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ORAL MANIFESTATIONS IN GENETIC SYNDROMES WITH MENTAL RETARDATION

INTRODUCTION

More than half of all cases of mental retardation (MR) are due to unknown causes. However, within the cases with recognizable genetic etiologies for MR, one can begin to appreciate the wide range of dental manifestations which can be present in a given syndrome. This manual is not intended to be a comprehensive review of all genetic and other associated syndromes involving MR. Instead, it is intended to be a user friendly outline of the most commonly encountered conditions within an institutional environment, and those frequently associated with oral manifestations. It is arranged in five sections relative to the etiology of the disorder.

- I. Chromosomal
- II. Single gene
 - A. Autosomal dominant
 - B. Autosomal recessive
 - C. Sex-linked
- III. Multifactorial
- IV. Environmental
- V. Sporadic and Unknown

CHROMOSOMAL SYNDROMES

4p Deletion (Wolf-Hirschhorn Syndrome)

Oral features:

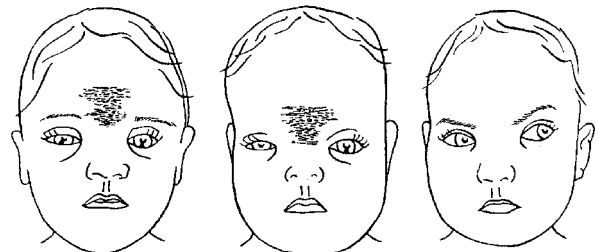
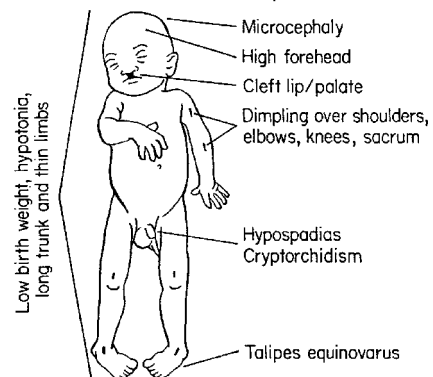
- cleft palate (40%) and cleft lip and palate (10%)
- micrognathia
- short philtrum
- down turned corners of mouth

General features:

- R microcephaly
- R wide nasal bridge
- R eye anomalies

4p Deletion (Wolf-Hirschhorn Syndrome)

MR + / all



Hemangioma on forehead, ptosis, epicanthal folds, hypertelorism, iris coloboma, short, deep philtrum, small downcurved mouth and short neck

5p Deletion (Cri-du-Chat Syndrome)

Oral features:

- micrognathia
- malocclusion - common, especially overjet
- cleft lip and palate

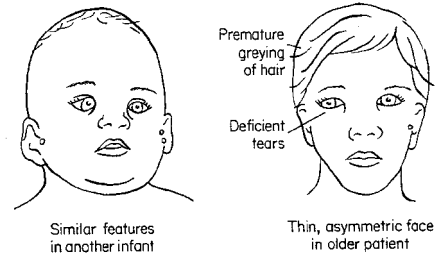
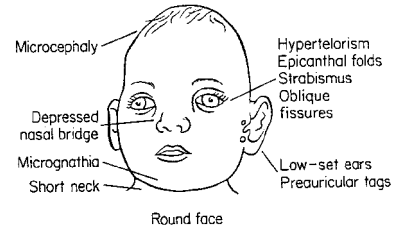
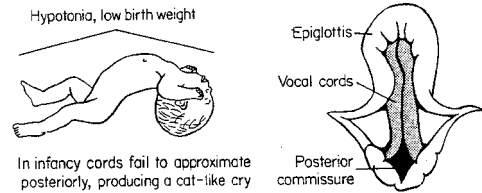
General features:

- R cat-like cry
- R microcephaly
- R round face
- R epicanthal folds
- R low set ears
- R severe psychomotor problems
- R mental retardation
- R hypotonia
- R hypertelorism
- R majority die in early childhood
- R some survive to adulthood (IQ<20)
- R incidence 1:20,000 to 50,000

Note: This syndrome is seen in about 1% of institutionalized patients.

5p Deletion (Cri-du-Chat Syndrome)

MR + / all



Trisomy 13 (Patau Syndrome)

Oral features:

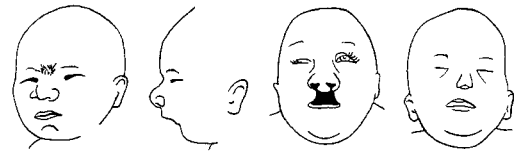
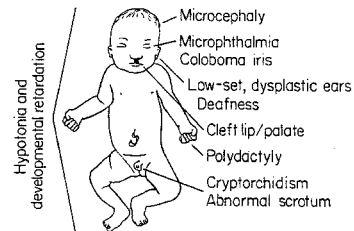
- cleft lip/palate (also premaxillary agenesis)
- micrognathia
- 45% die during first month, 86% during first year

General features:

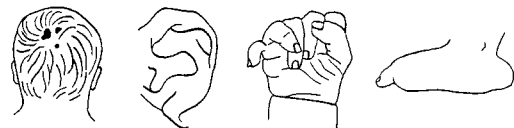
- R microcephaly/sloping forehead
- R microphthalmia/eye anomalies
- R postaxial polydactyly

Trisomy 13 (Patau Syndrome)

MR + / all



Facies with hemangioma on forehead, sparse/absent eyebrows, micrognathia, sloping forehead, low-set ears, microphthalmia, anophthalmia and single nares



Posterior scalp lesions, dysplastic ear, postaxial polydactyly, clenched hand, narrow nails and prominent heel

15q Deletion (Prader-Willi Syndrome)

Oral features:

- dental caries
- enamel hypoplasia
- malocclusion
- heavy calculus
- decreased salivation
- viscous, bubbly saliva
- gingivitis
- fish-like mouth with triangular shaped upper lip
- microdontia
- thick saliva at edges of mouth
- arched palate

General features:

- R hypotonia
- R obesity (complicates sedation)
- R small hands/feet
- R diminished fetal activity
- R severe skin picking behavior
- R hypopigmentation (75% show this)
- R diabetes onset following puberty
- R death usually due to diabetes or cardiac failure
- R hypogonadism
- R tend to be relatively insensitive to pain

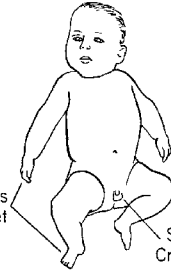
Note: This patient will benefit from regular prophylaxis.

15q Deletion (Prader Willi Syndrome)

MR + / most



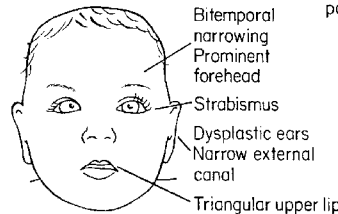
Hypotonia, poor sucking and swallowing reflexes at birth



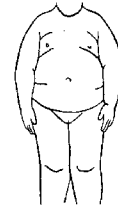
Small hands and feet

Small penis
Cryptorchidism

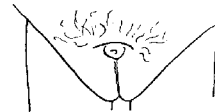
After first year polyphagia with obesity



Fat face with micrognathia



Short stature, decreased muscle with increased adipose tissue



Atrophic scrotum, small testes and penis

15q Deletion (Angelman Syndrome or Happy Puppet Syndrome)

Oral features:

- protruding tongue with drooling
 - mandibular prognathism
 - macrostomia
 - teeth widely spaced (probably due to tongue pressure)
- (No Illustration available)

Trisomy 18 (Edwards Syndrome)

Oral features:

- small mouth
- narrow palate
- micrognathia

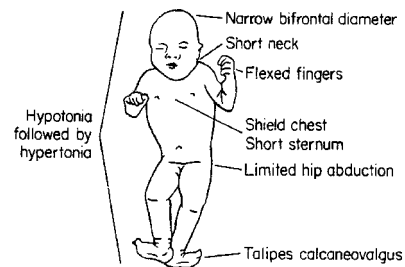
General features:

- R dolichocephaly/prominent occiput
- R short palpebral fissures/eye anomalies
- R overlapping fingers
- R rocker bottom feet

Note: Median life expectancy is 5 days.

Trisomy 18 (Edwards Syndrome)

MR + / all



Hypotonia followed by hypertonia



Small mouth
Short upper lip
Micrognathia

Low-set, dysplastic ears

Prominent occiput

Microphthalmia, epicanthal folds, short palpebral fissures, corneal opacities are common eye findings



Clenched hand, overlapping index fingers, short hallux dorsiflexed and prominent heel

Trisomy 21 (Down Syndrome) (See Module 3)

Oral features:

- enamel hypoplasia (primary dentition)
- periodontal disease (90%) and NUG (30%)
- narrow and short palate
- frontanels are large (third frontanel)
- frontanel and sphenoid sinuses are absent
- maxillary sinuses are hypoplastic
- irregular and delayed dental eruption
- missing teeth (3rd M, 2nd M, Lat. Incisors)
- decreased calculus formation
- caries rate is low
- hypodontia
- taurodontism
- malocclusion (crossbites and openbites common)
- parotid salivary flow rate is decreased and increase Na⁺, Ca²⁺, HCO₃⁻, and uric acid
- cardiac defect may be present requiring SBE prophylaxis
- lingual papillae are large
- protruding, large and fissured tongue
- lips are broad, irregular fissured, and dry
- open mouth

General features:

- R** brachycephaly/flat occiput
 - R** upward slanting palpebral fissures/epicanthal folds
 - R** brachydactyly/clinodactyly/simian crease
- Note: Atlantoaxial instability (10-20%) and occipitoatlantal instability is seen in these patients. These features are important particularly in relation to cervical extension during OR procedures or during full restraint procedures.

45,X (Turner Syndrome)

Oral features:

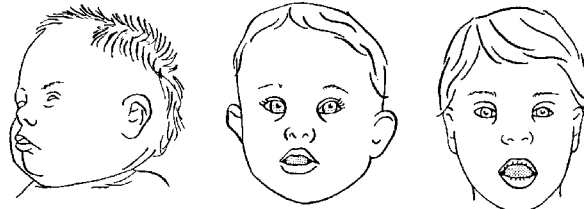
- micrognathia
- premature eruption of permanent molars
- high arched palate
- malocclusion

General features:

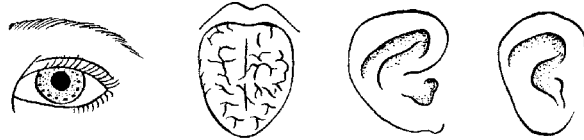
- R** excess skin neck
 - R** lymphedema hands/feet
 - R** shield chest/widely spaced nipples
 - R** hyperconvex, deep-set nails
- Note: Syndrome is seen only in females and usually not accompanied by mental retardation.

Trisomy 21 (Down Syndrome)

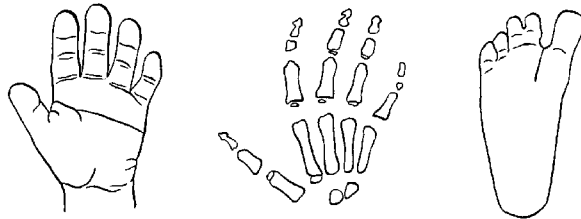
MR + / all



Short neck with skin folds, brachycephaly, flat face, protruding tongue, epicanthal folds, straight hair and small, low-set ears



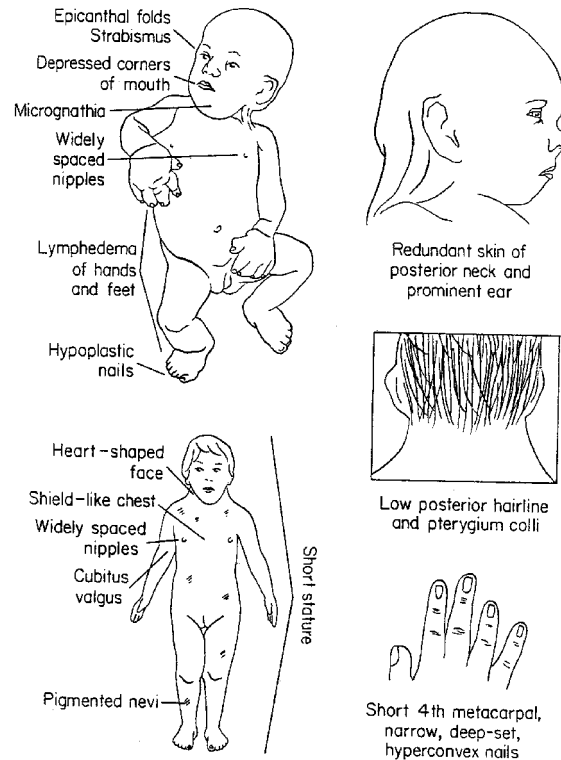
Epicanthal fold, Brushfield spots in iris, fissured tongue, angular overlapping helix, prominent antihelix and small ear lobes



Brachydactyly, clinodactyly, simian crease, small middle phalanx 2nd and 5th digits, syndactyly toes 2-3, wide space and furrow between toes 1-2

45 X (Turner Syndrome)

MR -



47,XXY (Klinefelter Syndrome)

Oral features:

- maxillary and mandibular prognathism (mandibular prognathism is more common)
- permanent tooth crowns larger than usual
- taurodontism
- no recognizable features apparent at birth (no illustration available)

Note: Syndrome seen only in males and often not accompanied by mental retardation.

47,XYY (XYY Syndrome)

Oral features:

- deciduous and permanent teeth larger than average
- shovel shaped lateral incisors
- no recognizable features at birth (no illustration available)

Note: Syndrome seen only in males and not usually accompanied by mental retardation.

SINGLE GENE SYNDROMES

Autosomal Dominant

Neurofibromatosis (NF-I)

Oral features:

- enlarged fungiform papilla
- oral neurofibromas (tumors may produce macroglossia)
- hyperplasia of soft and oral tissues associated with bony hypoplasia
- malpositioned teeth
- intrabony lesions
- wide inferior alveolar canals

General features:

- R multiple cafe-au-lait spots
- R axillary/intertriginous freckling
- R multiple neurofibromas
- R bony abnormalities

Tuberous Sclerosis

Oral features:

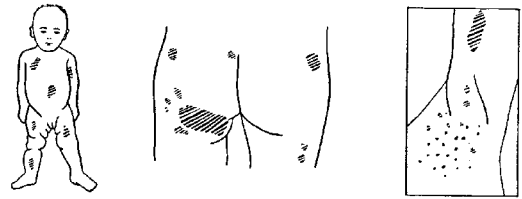
- pitted enamel
- gingival fibromas
- gingival hyperplasia (secondary to Dilantin therapy for seizures)

General features:

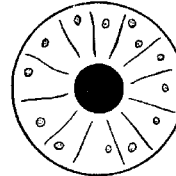
- R angiofibromas about face
- R white leaf-shaped skin lesions
- R subungual fibromas
- R seizures

Neurofibromatosis (NF-)

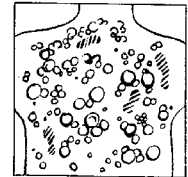
MR + / few



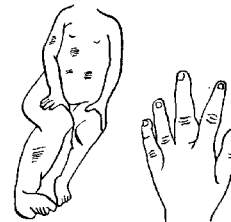
Six or more cafe-au-lait spots at birth and axillary freckling in a child



Lisch nodules in iris



Multiple neurofibromas about back



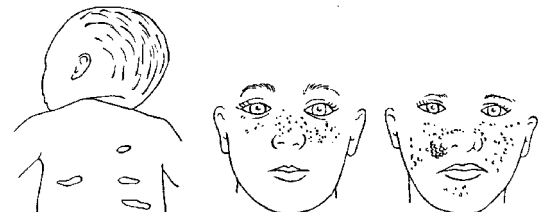
Hypertrophy of leg and fingers, pseudoarthrosis of tibia



Unilateral neurofibromatosis

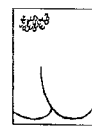
Tuberous Sclerosis

MR + / most

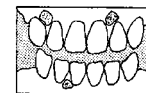


Multiple white macules on back of infant

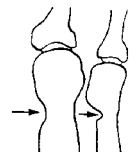
Angiofibromas about nasolabial folds



Shagreen patch lumbosacral area



Gingival fibromas



Lytic lesions of hand bones



Subungual fibromas about toes and finger



CT scan with periventricular calcifications

Apert and Carpenter Syndromes

Oral features:

- highly arched palate with lateral swellings
- clefting of soft palate (30%)
- severe malocclusion, especially Class III
- very crowded dentition
- anterior open bite
- midface hypoplasia
- relative mandibular prognathism
- delayed dental eruption
- reduced nasopharyngeal dimension

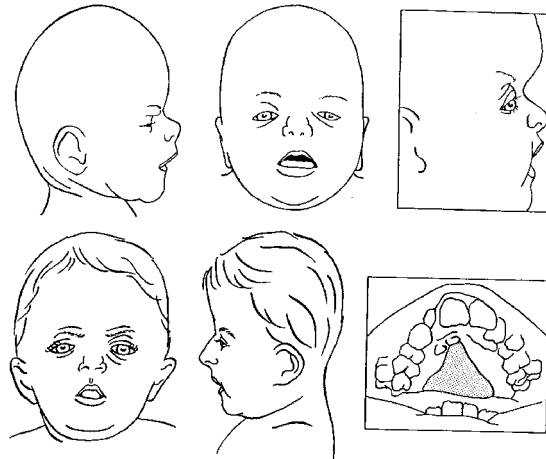
General features:

Apert Syndrome

- R turribrachycephaly
- R syndactyly of hands and feet
- R Craniosynostosis
- R Flat occiput
- R Steep forehead
- R middle third of face is retruded and hypoplastic
- R proptosis
- R mongoloid slanting (downward) of palpebral fissure
- R low set ears
- R hypertelorism

Apert Syndrome

MR + / most



Turribrachycephaly with high, steep, flat frontal bones, small pinched nose, strabismus, proptosis of eyes, downward slant to fissures, flat midface-narrow, high arched palate with dental malocclusion



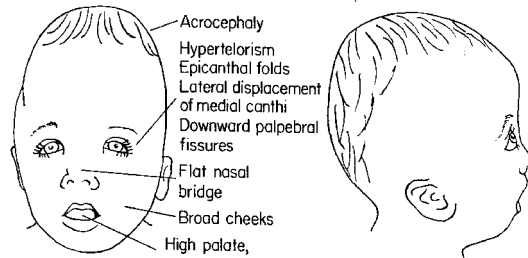
Varying degrees of syndactyly of fingers and toes

Carpenter Syndrome

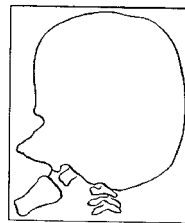
MR + / most

Carpenter Syndrome

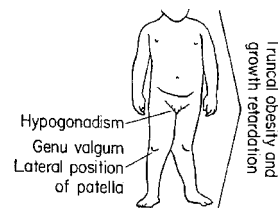
- R acrocephaly
- R flat nasal bridge
- R syndactyly fingers/polysyndactyly toes
- R obesity



Note low-set ears and micrognathia



Acrocephaly with high, straight forehead



Truncal obesity and growth retardation
Hypogonadism
Genu valgum
Lateral position of patella



Pes varus, broad hallux, polydactyly, syndactyly and brachydactyly

Stickler Syndrome

MR + / few

Stickler Syndrome

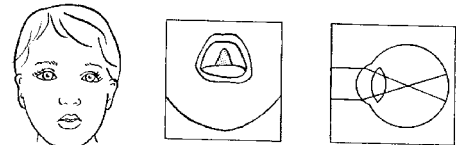
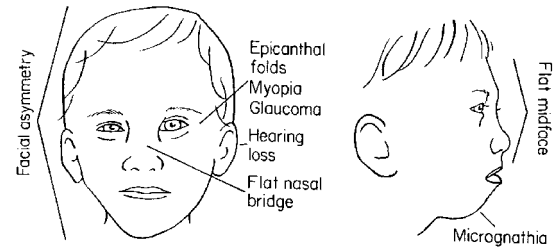
Oral features:

- midface hypoplasia, short maxilla
- long philtrum
- cleft palate
- abnormal mobility of soft palate

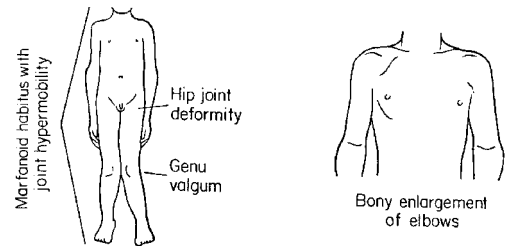
General features:

- R enlarged joints
- R myopia/retinal detachment
- R flat midface

Note: Approximately 30% of patients with Robin sequence have Stickler syndrome.



Rounded faces with asymmetry, cleft palate, myopia prone to retinal detachment



Autosomal Recessive

Hurler Syndrome (MPS I)

(Mucopolysaccharidosis I-H)

Oral features:

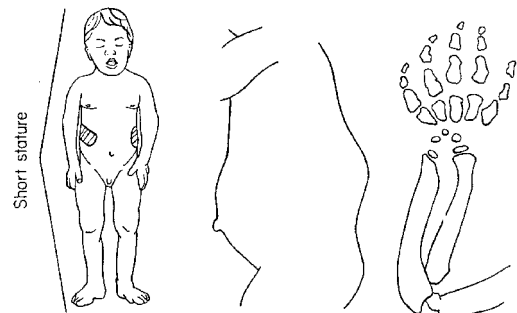
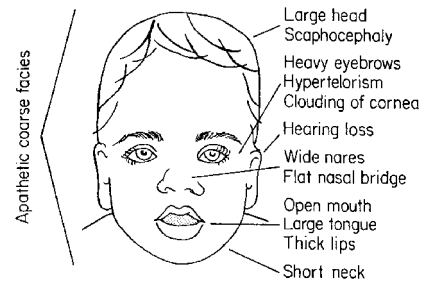
- flattened philtrum
- lip patulous
- widely spaced teeth
- lip and tongue enlargement after 5 years old
- incisors somewhat conical
- anterior open bite due to macroglossia
- ectopic molars in mandibular rami
- delayed eruption
- TMJ may exhibit limited motion
- dentigerous cysts
- airway obstruction
- sleep apnea reported
- mandible short and broad
- wide bigonial distance

General features:

- R macrocephaly/frontal bossing
- R coarse facies/thick ear lobes and lips
- R hepatosplenomegaly
- R chest and spine deformities

Hurler Syndrome (MPS I) (Mucopolysaccharidosis I-H)

MR + all



Protuberant abdomen with hepatosplenomegaly, umbilical hernia, gibbus deformity, joint limitation, claw-hand deformity and long bones are broad and short

EEC (Ectrodactyly-Ectodermal Dysplasia-Clefting Syndrome)

MR + / few

EEC (Ectrodactyly-Ectodermal Dysplasia-Clefting Syndrome)

Oral features:

- cleft palate alone in 10%
- cleft lip or palate (often bilateral in 75%)
- hypodontia, microdontia, anodontia
- conical teeth
- enamel dysplasia
- xerostomia contributing to high caries rate with dry granulosomatous lesions on lips
- parotid duct atresia

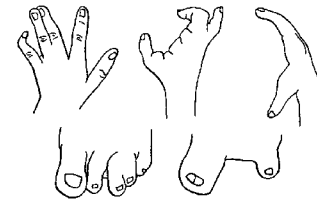
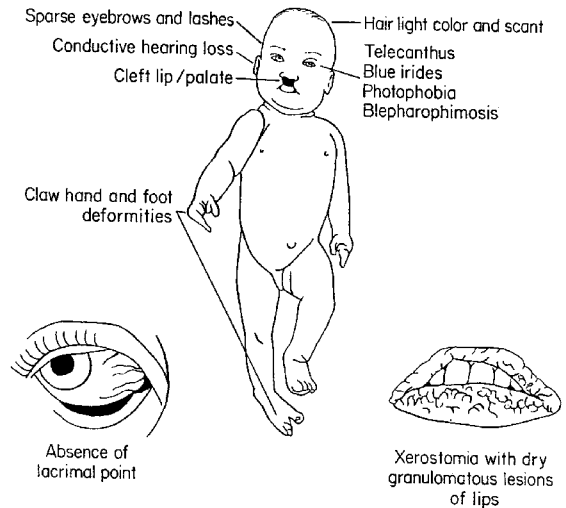
General features:

R sparse eyebrows/lashes

R scant scalp hair

R ectrodactyly

Note: Candidal cheilitis and candidal perleche have been described.



Syndactyly, clinodactyly, ectrodactyly and nail dystrophy of hands and feet

Coffin-Lowry Syndrome

MR + / all in males; / Most in females

Sex-linked

Coffin-Lowry Syndrome

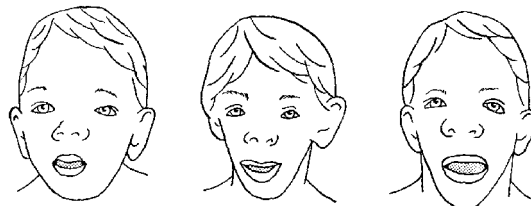
Oral features:

- mouth usually held open
- full thick lips with pouty lower lip
- relative mandibular prognathism due to hypoplastic maxilla
- malocclusion with overjet and/or overbite
- hypodontia of lower permanent teeth
- increased incidence of periodontal disease
- hypoplastic teeth

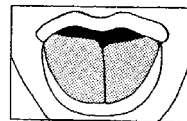
General features:

R square forehead/bitemporal narrowing

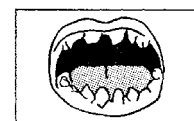
R prominent ears



Progressive coarsening of facial features with hypertelorism, downward slant of palpebral fissures, broad nasal bridge, thick nasal septum, anteverted nares, thick lips, pouting lower lip, prominent chin and large protruding ears



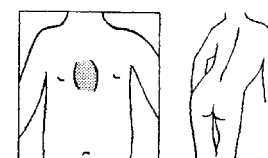
Deep tongue groove



Hypodontia and conical crowned incisors



Hyperextensible fingers, hand broad and thick, fingers taper distally



Pectus excavatum and kyphoscoliosis

Lesch-Nyhan Syndrome (See Module 16)

Oral features:

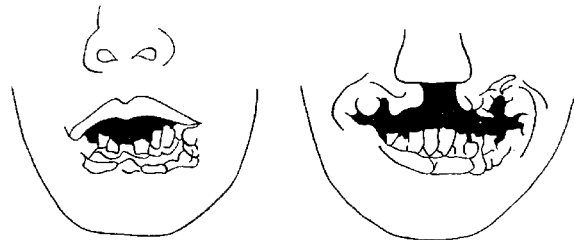
- self mutilating behavior usually involving the lips and fingers

General features:

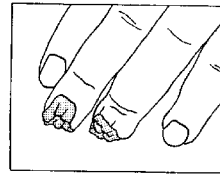
- R tophi about ears
- R choreoathetosis
- R spasticity
- R mental retardation
- R uric acid urinary stones
- R hyperurucemia

Lesch-Nyhan Syndrome

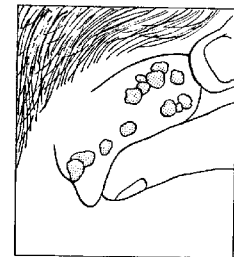
MR + / all



Varying degrees of self-mutilation of lips



Self-mutilation of fingers



Tophi on pinna

Hunter Syndrome (MPS II)
(Mucopolysaccharidosis II)

Oral features:

- widely spaced teeth
- tongue is enlarged
- neck is short

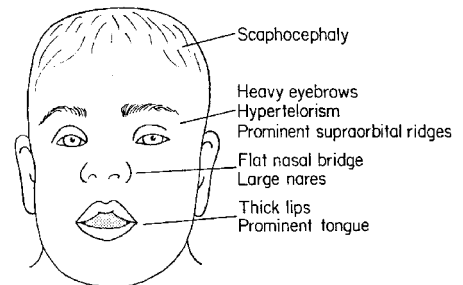
General features:

- R coarse facies/thick lips and nose
- R thick, hirsute skin
- R joint-stiffness/claw hand deformity
- R hepatosplenomegaly

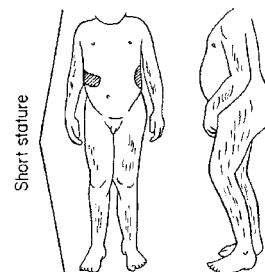
Note: Findings are similar to those in MPS I.

Hunter Syndrome
(MPS II) (Mucopolysaccharidosis II)

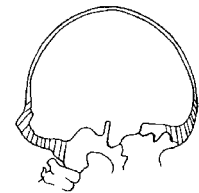
MR + / all, severe form



Coarse facies, short neck and progressive deafness



Hepatosplenomegaly, hypertrichosis, claw hand, stiff joints, pes cavus



Large sella turcica, frontal and occipital hyperostosis

Fragile X Syndrome

Oral features:

- high and narrow palate
- lateral palatine ridges prominent
- mandible also becomes particularly prominent in some males after age 20
- cross bite and open bite are common

General features:

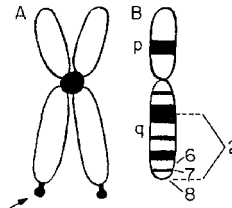
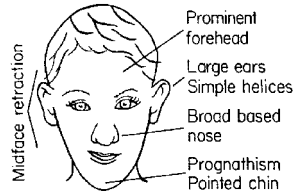
- R** prominent forehead
- R** large, simple ears
- R** prognathism (prominent symphysis rather than true prognathism)
- R** moderate to severe mental retardation
- R** high pitched jocular speech
- R** large testes after puberty
- R** deterioration of IQ with age

Note: There is a high incidence of MVP with mild aortic dilation requiring SBE prophylaxis.

Fragile X Syndrome

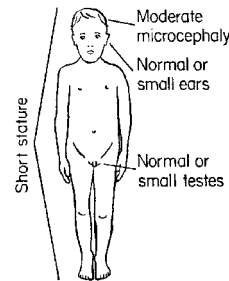
MR + / all

MACRO-ORCHIDISM/FRAGILE X SYNDROME



A) fragile X-chromosome (arrow)
B) fragile site at Xq28

RENPENNING SYNDROME



ALLEN-HERNDON-DUDLEY SYNDROME



Riley-Day type, Familial Dysautonomia

MR -

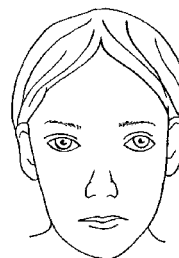
Riley-Day type, Familial Dysautonomia

Oral features:

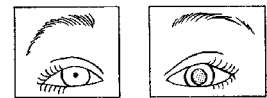
- characteristic slit-like mouth
- absent fungiform papillae
- dental cavities infrequent
- excessive drooling and diminished gag reflex

General features:

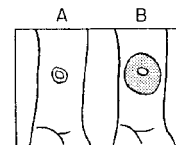
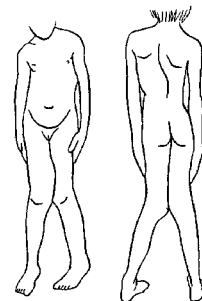
- R** hypotonia
- R** indifference to pain/decreased tearing
- R** absent or hypoplastic fungiform papillae
- R** thin, sad face/transverse mouth
- R** self mutilation



Normal tongue with fungiform papillae—tongue of a patient with absence of fungiform papillae, note drooling



2.5% methacholine in right eye producing miosis—no effect on a normal pupil



Histamine test in a patient (A), wheal but no large flare as in the normal (B)

MULTIFACTORIAL SYNDROMES

NTD (Neural Tube Defect)

Anencephaly (universally lethal)

Spina Bifida

Oral features:

- dental manifestations can include a high and narrow palate
- avoid latex dental devices

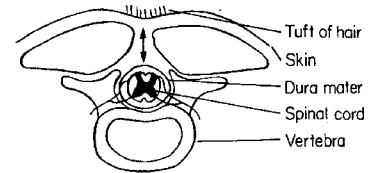
General features:

- R variable degrees of lower extremity paralysis
- R bladder and bowel incontinence
- R secondary hydrocephaly sometimes present
- R sacral hairy patch or dimple
- R diastematomyelia
- R intradural/extradural lipoma
- R caudal regression syndrome
- R sacral agenesis
- R intellect may or may not be affected depending on presence or absence of hydrocephaly
- R anatomical defect in region of the vertebral column

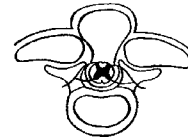
Note: Individuals with spina bifida are frequently allergic to latex products.

NTD (Neural Tube Defect)

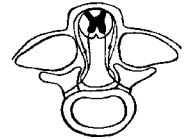
MR + / few



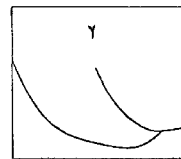
Spina bifida occulta (arrow shows defect)



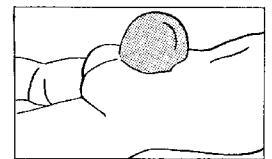
Meningocele



Meningomyelocele



Skin dimple over site of spina bifida



Meningomyelocele-lumbosacral area



Meningomyelocele-dorsolumbar, lumbar and thoracolumbar

Robin Sequence (Pierre Robin Syndrome)

Oral features:

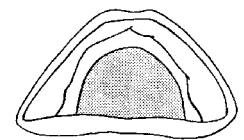
- severe microretrognathia, hypoplastic mandible
- glossoptosis
- U-shaped cleft palate
- respiratory difficulties, secondary to micrognathia and glossoptosis and narrow upper airway
- impaction of tongue in palatal cleft can result in asphyxia

Robin Sequence (Pierre Robin Syndrome)

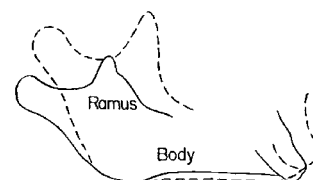
MR + / few



Severe micrognathia and low-set dysplastic ear



U-shaped cleft palate



Normal mandible broken line, Robin solid line - note difference in height of ramus, length of body and inclination of ramus to body

ENVIRONMENTAL SYNDROMES

FAS (Fetal Alcohol Syndrome)

Oral features:

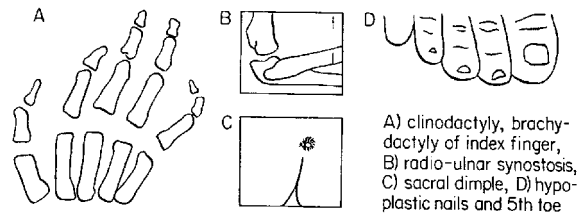
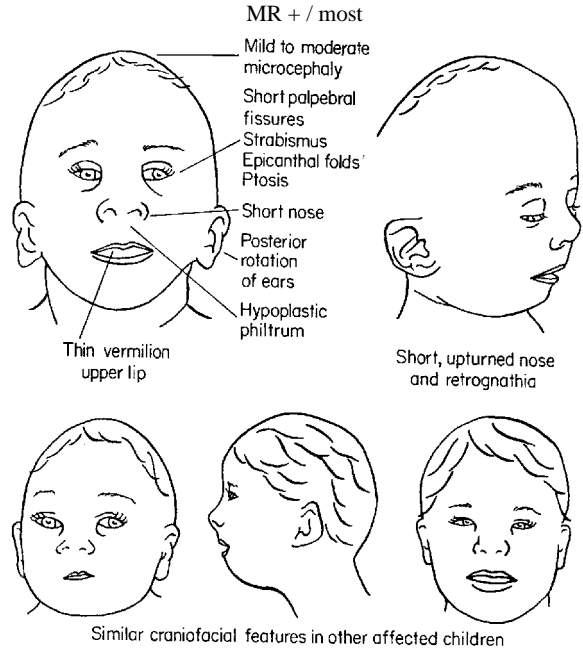
- smooth philtrum
- thin upper lip
- mild micrognathia
- hypoplastic mandible
- cleft palate in some cases

General features:

- R** irritable infant
- R** growth (length) retardation
- R** narrow palpebral fissures

Note: Syndromes caused by congenital infections are not usually associated with specific outstanding pathognomonic dental manifestations. They are not always associated with MR.

FAS (Fetal Alcohol Syndrome)



SPORADIC AND UNKNOWN

ETIOLOGY SYNDROMES

Beckwith-Wiedemann Syndrome

Oral features:

- macroglossia
- malocclusion, anterior open bite
- widely spaced teeth likely due to macroglossia
- speech problems due to macroglossia

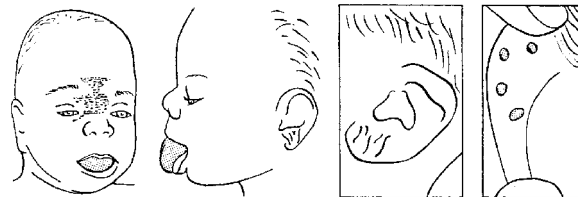
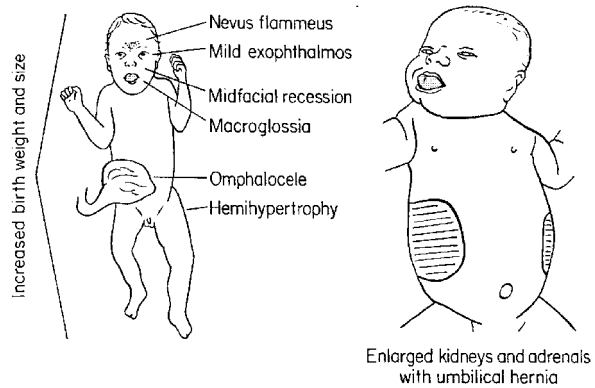
General features:

- R** omphalocele
- R** earlobe grooves/depressions on posterior helix
- R** visceromegaly

Note: Glossectomy may be beneficial for speech and tongue size

Beckwith-Wiedemann Syndrome

MR + / most



Nevus flammeus, macroglossia, linear fissures on earlobe and punched-out depressions on posterior pinna



Sturge-Weber Syndrome

MR + / some

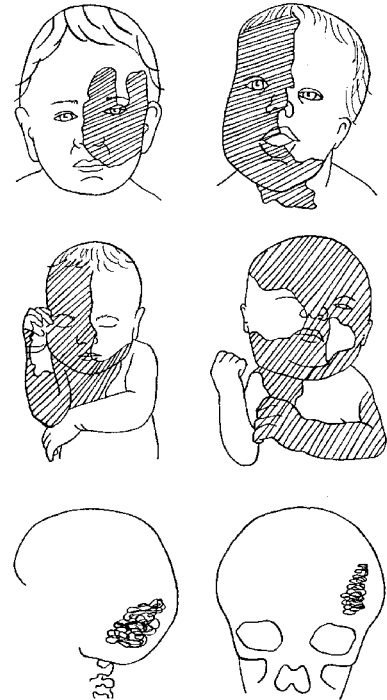
Sturge-Weber Syndrome

Oral features:

- hemangioma formation may require patient to be treated in the OR
- gingival hyperplasia — vascular type should be distinguished from fibrous
- bleeding problems for extractions
- eruption of teeth on affected side more advanced than on unaffected side
- angiomatosis involving buccal mucosa and lips
- gingival vascular lesions, when present, may range from mild enlargements to extreme overgrowth making closure impossible
- macrocheila (large lips)
- hemihypertrophy of tongue
- hypertrophy or hypotrophy of alveolar process
- ipsilateral premature, delayed or normal eruption leading to malocclusion

General features:

- R unilateral angiomatosis about face, chest, and upper extremity
- R seizures
- R glaucoma on ipsilateral side
- R hemiplegia
- R ipsilateral calcification of cerebral cortex
- R macrocephaly and skull asymmetry noted



Nevus flammeus mainly unilateral about face with facial hypertrophy, eye, forehead, upper chest and extremity—areas of intracranial calcification

Williams Syndrome

MR + / all

Williams Syndrome

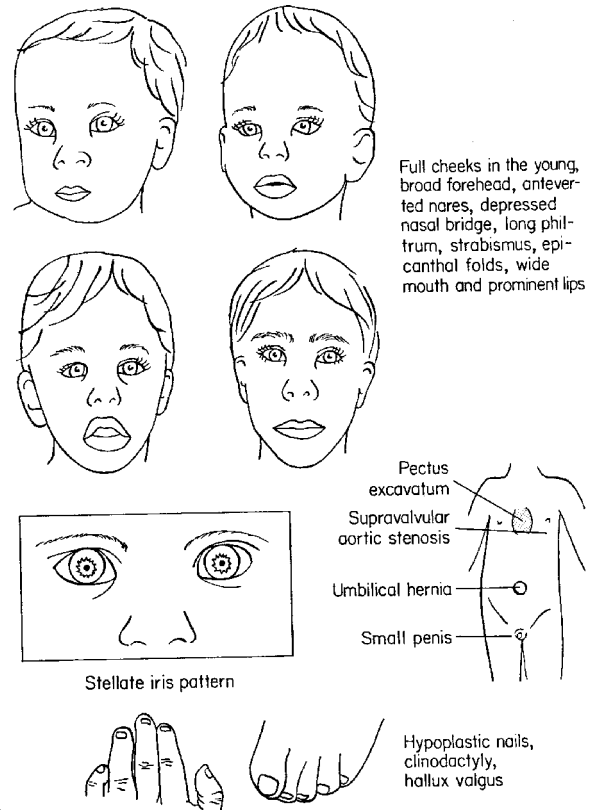
Oral features:

- long philtrum
- thick, wide lips
- maxillary arch too broad for mandibular arch
- hypodontia
- microdontia
- open mouth
- dens invaginatus
- mild macrognathia
- delayed mineralization of teeth
- prominent and accessory labial frenula
- hypoplastic bud shaped primary and permanent molars

General features:

- R full, dependent cheeks
- R heart murmur

Note: Heart murmur (supravalvular stenosis) may be present requiring SBE prophylaxis.



Full cheeks in the young, broad forehead, anteverted nares, depressed nasal bridge, long philtrum, strabismus, epicanthal folds, wide mouth and prominent lips

Stellate iris pattern

Pectus excavatum
Supravalvular aortic stenosis
Umbilical hernia
Small penis

Hypoplastic nails, clinodactyly, hallux valgus

Cornelia de Lange Syndrome

MR + /all

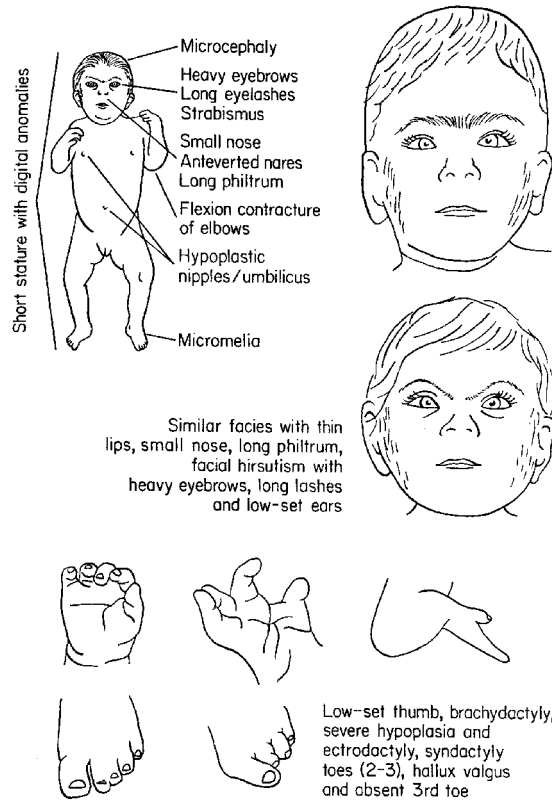
Cornelia de Lange Syndrome

Oral features:

- micrognathia
- down turned, thin lips
- cleft palate in 20%
- delayed tooth eruption and widely spaced teeth
- microdontia
- often severe dental behavior problems

General features:

- R** microbrachycephaly
- R** synophrys/long eyelashes
- R** hand and limb anomalies
- R** delayed bone age
- R** abnormal speech pattern
- R** frequently do not express facial expression
- R** low hairline on forehead and neck
- R** short, thick neck
- R** pigmented nevi on skin



Silver Syndrome (Russell-Silver Syndrome)

MR + / few, mild

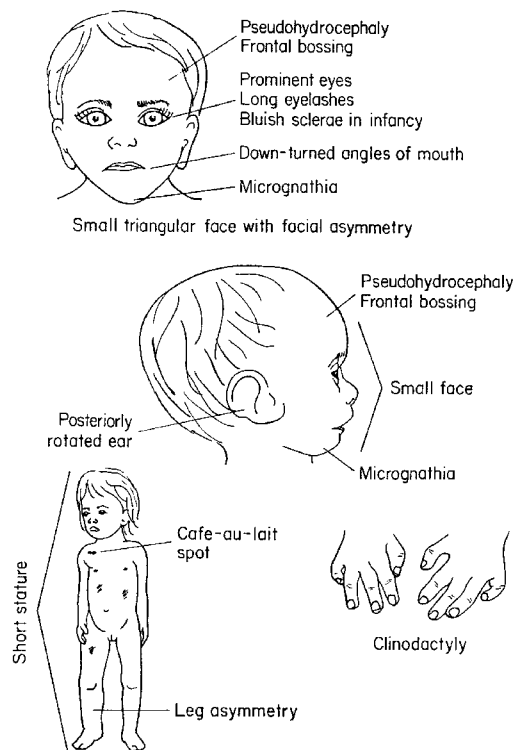
Silver Syndrome (Russell-Silver Syndrome)

Oral features:

- high and narrow palate
- teeth are crowded
- micrognathia

General features:

- R** asymmetry body/limb/face
- R** triangular face/long eyelashes
- R** frontal bossing



GLOSSARY

acrocephaly (oxycephaly) - a type of craniosynostosis in which there is a premature closure of the lambdoid and coronal sutures, resulting in an abnormally high, peaked, or conically shaped skull, "tower skull."

angioma - a swelling or tumor due to proliferation with or without dilation of the blood vessels (hemangioma) or lymphatics (lymphangioma).

angiomatosis - a condition characterized by multiple angiomas.

anophthalmia - congenital absence of one or both eyes.

athetosis - a condition in which there is a constant succession of slow, writhing, involuntary movements of flexion, extension, pronation, and supination of the fingers and hands, and sometimes of the toes and feet.

atlantoaxial instability - malalignment of cervical vertebrae C-1 and C-2 (atlas and axis) in the neck; also occipitatlantal instability.

bigonial - distance between the angles of the mandible.

blepharophimosis - inability to open the eye to the normal extent.

brachydactyly - shortness of the fingers.

chorea - a disorder characterized by irregular, spasmodic, involuntary movements of the limbs or facial muscles.

choreoathetosis - abnormal movements of body of combined choreic and athetoid pattern.

clinodactyly - permanent deflection of one or more fingers.

coloboma - any defect, congenital, pathologic, or artificial, especially of the eye.

cryptorchidism - having testes which have not descended into the scrotum.

cubitus valgus - deviation of the extended forearm to the outer (radial) side of the axis of the limb.

dolichocephaly - the condition of having a disproportionately long head.

ectrodactyly - congenital absence of one or more fingers or toes.

exophthalmos - protrusion of the eyeballs.

genu valgum - a deformity marked by abduction of the leg in relation to the thigh, "knock-knee."

gibbus deformity - extreme kyphosis, hump or hunch.

glossoptosis - downward displacement of the tongue.

hallux - the great toe, "the big toe."

hallux valgus - displacement of the great toe toward other toes.

hypertelorism - abnormal distance between two paired organs, e.g. widely-spaced eyes.

hypertrichosis - excess hair growth.

hypospadias - a developmental anomaly characterized by a defect in the wall of the urethra so that the canal is open for a greater or lesser distance on the under surface of the penis.

kyphoscoliosis - kyphosis combined with scoliosis.

kyphosis - an abnormal curvature of the spine, with convexity backward (cyphosis).

meningocele - congenital hernia in which the meninges protrude through an opening of the skull or spinal column.

meningomyelocele - hernia of the spinal cord and membranes through a defect in the vertebral column.

microphthalmia - the presence of one or both eyeballs of abnormally small size.

miosis - contraction of the pupil.

nevus flammeus - port-wine mark or stain. A large nevus vascularis having a purplish color. It is usually found on the head and neck and persists throughout life.

omphalocele - congenital hernia of the navel.

patulous - patent, lying freely open.**perleche** - disorder marked by fissures and epithelial desquamation at corners of the mouth.

pes varus - talipes varus. A form of clubfoot in which the foot is bent or twisted inward.

phalanx - any one of the bones of the fingers or toes.

polydactyly - the presence of more than five digits on either hand or foot.

polyphagia - eating abnormally large amounts of food.

postaxial polydactyly - polydactyly occurring on the "little finger" side (most common type of polydactyly).

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