

Surface Evaluation for Minor Congenital Anomalies

Adapted from "The Physician's Guide to Caring for Children with Disabilities and Chronic Conditions" – Nickel, R.E. and Desch, L.W. (Editors). (2000). Paul H. Brookes Publishing Co.

Variation of normal: Feature occurring in >4% of population and has no cosmetic or functional significance to the individual.

Minor malformation: Feature occurring in <4% of population and has no cosmetic or functional significance to the individual.

Major malformation: Feature that has cosmetic or functional significance to the individual.

The purpose of this examination is to provide a structure for making skilled observations to assist with the identification of children with birth defect syndromes and genetic problems. This examination is best integrated in the general physical examination. When first learning to evaluate a child thoroughly for minor anomalies, however, it is useful to conduct the examination from start to finish to become comfortable with every aspect of the examination. Note that any newborn with two or more major defects (such as a congenital heart defect, cleft lip and palate or syndactyly) or three or more minor congenital anomalies may have a chromosomal disorder or birth defect syndrome.

Starting with the head and face, follow the sequence presented below. For each stage of the examination, a list of the most common anomalies is provided. Please remember to measure whatever can appropriately be measured (e.g., ear length, hand length, palpebral fissure length). Circle the anomalies that are present and describe or write in the measurements in the free space on the right. Please also describe any anomalies next to the appropriate category that are not listed on the form.

Craniofacial

Flat or prominent nasal bridge
Small mandible
Flat or prominent occiput
Metopic ridge
Large posterior fontanelle
Malar hypoplasia
Anteverted nose
Synophrys

Eyes

Epicanthal folds
Hypo- and hyper-telorism
Ptosis
Short palpebral fissures
Upward slant to palpebral fissures
Downward slant to palpebral fissures

Chest

Short sternum
Depressed sternum
Wide-set or high-located nipples
Shield chest

Abdominal/perineal

Diastasis recti (>3 cm)
Umbilical hernia
Inguinal hernia
Small testes
Hypospadias
Small or hypoplastic genitals

Hands

Single palmar crease
Other unusual crease pattern
Clinodactyly
Camptodactyly
Partial cutaneous syndactyly
Proximally placed thumb
Broad thumb
Duplication of thumbnail
Small or dysplastic nails
Overlapping fingers
Long fingers
Small or large hands
Short metacarpals

Ears

Preauricular tags or sinus
Large or small ears
Asymmetric size
Low-set ears
Posterior rotation (>20%)
Lack of usual fold of helix

Mouth

Bifid uvula
High-arched palate
Wide alveolar ridges
Large tongue
Thin upper lip
Flat philtrum

Skin/hair

Low hairline
Frontal upsweep/aberrant hair whorl
Alopecia of scalp
Extra posterior cervical skin
Large capillary hemangioma (Other than on posterior neck)
Café au lait spots
Hypopigmented macules
Deep sacral dimple
Aplasia cutis congenita

Feet

Syndactyly of toes
Overlapping toes
Wide gap ("sandal-gap") between toes
Prominent heel
Broad hallux
Hallux valgus
Hypoplastic nails
Duplication of nail (rudimentary polydactyly)

Reference: Jones, K.L. (Ed.). (2006). *Smith's Recognizable Patterns of Human Malformation* (6th ed.). Philadelphia: Elsevier Saunders

Normal standards:

Outer canthal distance: page 856
Inner canthal distance: page 857
Palpebral fissure length: page 858
Ear length: page 861
Total hand length: page 852
Palm length: page 853
Middle finger length: page 853
Foot length: page 855
Penile length: page 862

Notes (please review Jones, pp. 817-836):

1. Measure the outer and inner canthal distance with a plastic see-through ruler.
2. A flat nasal bridge and anteverted nose typically go together. Consider a nose anteverted if you can see straight into the nostrils when looking at the child from the front.
3. Ears are posteriorly rotated (slanted away from the eye) if there is a 15% slant away from the perpendicular (Jones, page 821).
4. A sacral dimple is considered deep if the bottom cannot be seen without considerable stretching. It should be distinguished from a pilonidal sinus.
5. Measure penile length by resting one end of a ruler on the pubic bone and stretching the penis as much as possible. Measure to the tip of the glans.
6. Hypoplastic testes refer to small size and/or abnormal consistency. Hypoplasia of the labia majora may give the impression of a large clitoris (Jones, page 825).
7. If the first metacarpal bone is short, the thumb will be proximally placed. If other metacarpals or metatarsals are short, the corresponding finger or toe will appear short. To check for a short metacarpal, have the child make a fist and check the knuckles. If a short metacarpal bone is present, the knuckle will be absent. A common example is relative shortness of the 4th or 5th metacarpal or metatarsal (Jones, page 822-823).
8. Dysplastic nails are spoon-shaped, ridged, or otherwise malformed nails. The nails generally reflect the size and shape of the underlying distal phalanx (Jones, page 823).
9. Partial syndactyly most commonly occurs between the 3rd and 4th fingers and 2nd and 3rd toes. Less than 25% syndactyly between the 2nd and 3rd toes is considered normal (Jones, page 824).