

### 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* (6<sup>th</sup> 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 4<sup>th</sup> or 5<sup>th</sup> 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 3<sup>rd</sup> and 4<sup>th</sup> fingers and 2<sup>nd</sup> and 3<sup>rd</sup> toes. Less than 25% syndactyly between the 2<sup>nd</sup> and 3<sup>rd</sup> toes is considered normal (Jones, page 824).