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)


**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. 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).
7. 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).
8. 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).