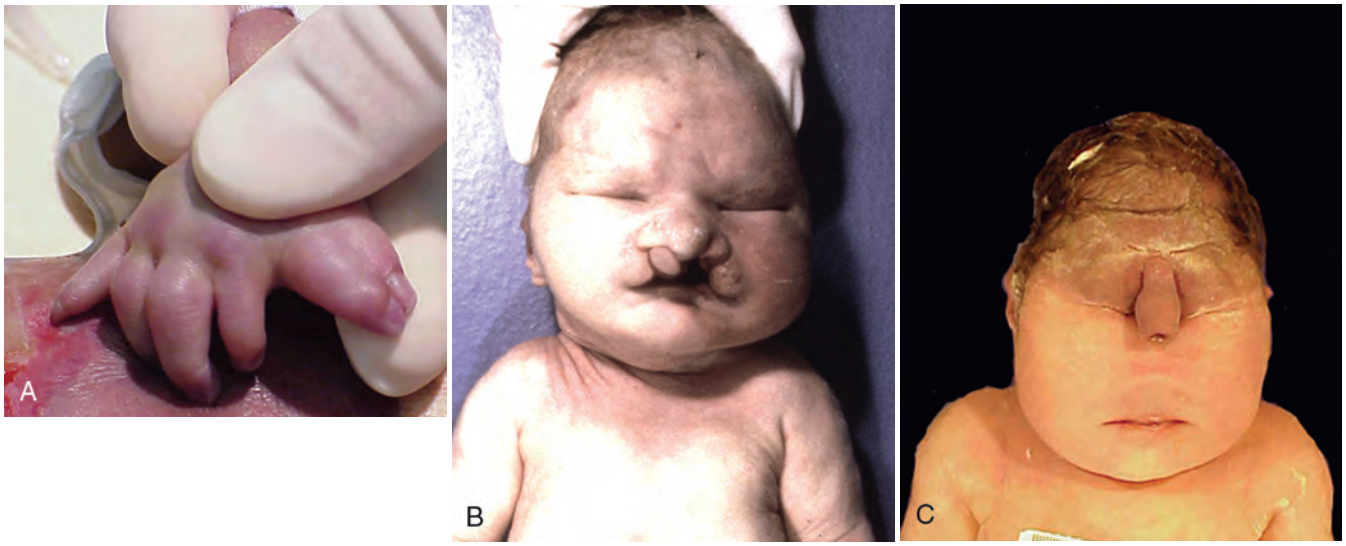

Figure 10-1 Examples of malformations. *Polydactyly* (one or more extra digits) and *syndactyly* (fusion of digits), both of which are illustrated in **A**, have little functional consequence when they occur in isolation. Similarly, *cleft lip* (**B**), with or without associated *cleft palate*, is compatible with life when it occurs as an isolated anomaly; in the present case, however, this neonate had an underlying *malformation syndrome* (trisomy 13) and died of severe cardiac defects. **C,** The stillbirth illustrated represents a severe and essentially lethal malformation, wherein the midface structures are fused or ill-formed; in almost all cases, this degree of external dysmorphogenesis is associated with severe internal anomalies such as maldevelopment of the brain and cardiac defects. (**A** and **C**, Courtesy Dr. Reade Quinton; **B**, Courtesy Dr. Beverly Rogers, Department of Pathology, University of Texas Southwestern Medical Center, Dallas, Texas.)

sets into motion secondary effects in other organs. A good example is the *oligohydramnios* (or *Potter*) *sequence* (Fig. 10-3). Oligohydramnios (decreased amniotic fluid) may be caused by a variety of unrelated maternal, placental, or fetal abnormalities. Causes of oligohydramnios include chronic leakage of amniotic fluid because of rupture of the amnion, uteroplacental insufficiency resulting from maternal hypertension or severe toxemia, and renal agenesis in the fetus (because fetal urine is a major constituent of amniotic fluid). The fetal compression associated with significant oligohydramnios, in turn, results in a classic phenotype in the newborn infant, including flattened facies and positional abnormalities of the hands and feet (Fig. 10-4). The hips may be dislocated. Growth of the chest wall and the contained lungs is also compromised so that the lungs are frequently hypoplastic, occasionally to the degree that they are the cause of fetal demise. Nodules in the amnion *(amnion nodosum)* are frequently present.

- A *malformation syndrome* is a constellation of congenital anomalies, believed to be pathologically related, that, in contrast to a sequence, cannot be explained on the basis of a single, localized, initiating defect. Syndromes are most often caused by a single etiologic agent, such as a viral infection or specific chromosomal abnormality, which simultaneously affects several tissues.

In addition to the aforementioned general definitions, a few organ-specific terms should be defined. Agenesis

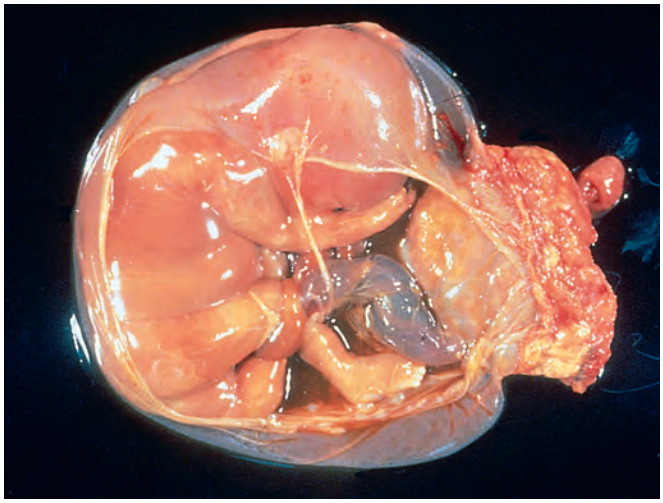

Figure 10-2 Disruption of morphogenesis by an amniotic band. Note the placenta at the right of the diagram and the band of amnion extending from the top portion of the amniotic sac to encircle the leg of the fetus. (Courtesy Dr. Theonia Boyd, Children's Hospital of Boston, Boston, Mass.)

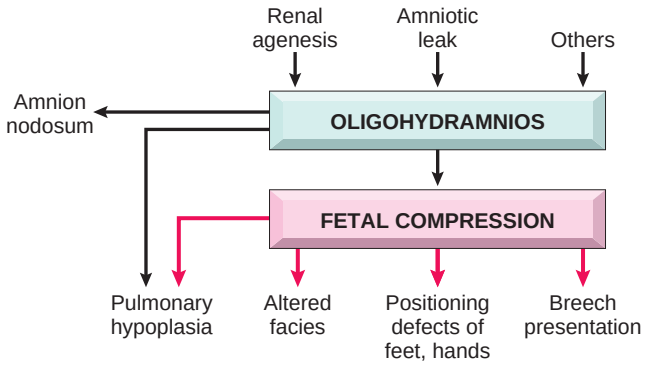

Figure 10-3 Schematic diagram of the pathogenesis of the oligohydramnios sequence.