The craniosynostoses are serious abnormalities of infancy and childhood. The term craniosynostosis (CS) indicates a cranial or craniofacial dysmorphism characterized by premature closure of one or more sutures of the cranial vault and/or base. This term was introduced by Sear in 1937 [1], but the recognition of these affections has been ascribed to Virchow [2], who first used the term CS. Virchow codified the general rules to explain cranial deformities, based on a concept of growth interruption of the skull perpendicular to the suture involved and consequent compensatory growth of the cranium parallel to the “pathological” suture. This concept, widely known as “Virchow’s law,” remains valid after 150 years, even though knowledge concerning the epidemiology, etiology, pathogenesis, and clinical/surgical therapy has dramatically changed and increased, especially during the last 20 years. Recently acquired genetic notions indicate that the basic concepts, as well as the medical-surgical approach to these pathologies, will radically change in the future.

The suspicion of CS is usually clinical, based on a cranial deformity. The diagnosis has always been almost exclusively radiological, and was recently revolutionized by the introduction of digital techniques, above all computed tomography with three-dimensional reconstruction (3DCT). The aim of imaging-based evaluation is to define the suture(s) involved, to image the deformity of the skull vault and base (required for planning surgical correction), and to exclude intracranial and/or brain alterations (which could be either a consequence of or associated with CS). The neuroimaging work-up is also important in monitoring the results of treatment and the “natural” evolution of the disease.

In this chapter, we will define the indications, applications, and limits of neuroimaging procedures in the diagnosis of CS. First, the epidemiology, etiopathogenesis, and classifications of CS will be delineated. Then, we will attempt to explain the evolution of neuroimaging as applied to CS, review all applica-
multiple imaging methods and, above all, define the rational indications for the various imaging procedures in terms of cost-effectiveness. In a more detailed discussion of 3DCT, we will then present the available techniques, their semeiotics, and the results obtainable, taking into account the most recent technological advances. The most common and significant clinical-radiological presentations of CS will then be illustrated in order to emphasize the optimal use of 3DCT and, when necessary, MRI. The last section will be dedicated to neuroimaging follow-up, especially after surgery.

30.2 Epidemiology

Generally, the deformities that result from CS are recognized in infancy. Reported incidence rates of all forms of CS range from 1:1,900 to 1:4,000 births, with an average incidence of approximately 1:2,000 births [3]. The prevalence of CS in the general population ranges from 34 to 48 per 100,000 live births, with syndromic cases being less common than nonsyndromic ones [4, 5]. CS is almost always present in children with Crouzon, Apert, or Pfeiffer syndromes [6, 7].

Several clusters of CS in Colorado areas in the 1980s greatly increased public awareness of the disease [8]. These cases were nonsyndromic and involved multiple sutures in patterns not previously described. The Center for Disease Control sponsored several large epidemiological studies in response to allegations that environmental factors (for example, water or radiation) were the cause. The results did not indicate a correlation with these factors, but did indicate a correlation between maternal behaviors and the closure of specific sutures. Another theory that was advanced, though unproven, was that raised awareness of this disease among local practitioners resulted in an increase of the frequency of diagnosis [8, 9].

In 1993, Kirby et al. [10] reported an apparent increase of CS in infants born in the York and Selby areas of Yorkshire between September, 1990 and November, 1992. In order to investigate this trend, they examined the Regional Information System data for Yorkshire from the previous 14 years and noted an exponential increase of admissions with an International Classification of Disease code which includes CS.

Since 1992, there has been an increase in the number of infants seen with deformational posterior plagiocephaly. The most likely explanations were the recommendation that infants slept in the supine position to decrease the risk of sudden infant death syndrome and the increased awareness of plagiocephaly among pediatricians and other primary care providers [11, 12].

30.3 Pathogenesis

Little is known about the pathogenesis of CS. The idiopathic primary form usually begins in utero or shortly after birth, although it is usually diagnosed in infancy due to the resulting skull deformities. One or more sutures may be affected. Genetics seems to play an important role; recently, the genes responsible for Apert, Crouzon, Pfeiffer, and Saethre-Chotzen syndromes have been located [13].

CS is considered to be a late developmental defect during embryogenesis (>17 mm crown-rump length) [14]. The frontal bone starts to ossify in a pair of bone centers (left and right), and each parietal bone in two fusing bone centers [15, 16].

Agensis of bone centers with subsequent agenesis of the involved bone and failure of bone centers to fuse where they normally do have been reported in literature [13, 17, 18].

Vermeij-Keers [14] postulated that CS could be caused by direct fusion of bone centers during embryogenesis. Adjacent bone centers are displaced toward the synostotic suture where they can undergo direct fusion. This malformation can occur unilaterally (e.g., unilateral coronal suture synostosis) and bilaterally (e.g., bilateral coronal suture synostosis in Apert syndrome). Severe CS, characterized by involvement of a larger number or even all cranial sutures, may impair brain growth and cause seizures, mental retardation, increased intracranial pressure, and visual loss [19–21]. The growing brain generates forces that cause compensatory growth at the sutures causing them to remain open, which results in deformity of the calvarium.

Some forms of CS have been linked to teratogens such as retinoic, diphenylhydantoin, and valproic acids [22].

Compression of the skull during the last months of pregnancy can favor the onset of the disease [22]. Moss [23] proposed that the cranial vault should be viewed as consisting of independent but coordinated functional components which are based on the major functions of the head, such as sight, speech, etc. The various bones that comprise the skull should be viewed as components of their functional groups. Moss also demonstrated that the growth of the neural