Introduction

Two terms are used currently to describe congenital defects in the cerebral hemisphere. The term “porencephaly”, originally coined by Heschl, was defined as follows: “the cerebral substance is lacking through the entire thickness of brain and so, if one disregards the purely membranous parts (which fill the defect), there is a
canal through the brain which begins on the outer surface of the brain and ends in the cerebral ventricles” (quoted from [1]). This term is now used to describe circumscribed hemispheric defects originating during fetal life and antedating the acquisition of a mature astroglial response or completion of convolutional development [2]. The other term “schizencephaly” was proposed by Yakovlev, who described two types. While the first type, “clefts with fused lips” was a distinct entity, the second type “clefts with lips separated” fits the definition of Heschl’s “porencephaly” [3]. Barth proposed that Heschl’s porencephaly and Yakovlev’s schizencephaly be considered parts of a spectrum of fetal disruption. At one end of this spectrum are the post-migration accidents resulting in lesions without gray matter lining the clefts or an associated malformation of cortical development. In the middle are the full-thickness defects, lined by gray matter, with adjacent polymicrogyria. At the other end are the cases with full-thickness defects with abnormal neocortex bordering the external part of the cleft and heterotopic gray-matter masses on the inside right up to the ventricular end of the cleft [3]. In some textbooks of neuropathology, clefts without communication between the cerebral surface and the ventricles are also included at the far end of this spectrum [2, 4, 5, 6]. We have chosen to use the term “schizencephaly” to designate the second and third types of the spectrum that Barth described. Our patients include only those meeting this definition; those patients without gray matter lining the clefts, presumably due to brain injuries after completion of neuronal migration, and those with clefts not in communication with the ventricles, were excluded.

Although there is an abundant literature describing the macroscopic pathological findings in schizencephaly, the findings in these autopsy cases are likely biased towards the more severe anomalies, because patients with milder anomalies seldom have autopsies. In contrast to autopsy, non-invasive imaging studies are performed on patients with milder, as well as severe, anomalies. It is important to know the full morphological spectrum of features and associated anomalies of schizencephaly, including those with minor anomalies, because this knowledge is helpful in establishing the imaging diagnosis. Previous MRI studies of schizencephaly have been sporadic, and few studies have carefully described the morphological variations in affected patients [7, 8]. This retrospective MR analysis of schizencephaly was performed in order to confirm the spectrum and the prevalence of macroscopic findings of schizencephaly in the clinical population, paying special attention to concomitant anomalies.

Methods

A review of the teaching files and radiological information system at our institution yielded 44 patients with MRI findings suggest-