ABSTRACT. Smith-Magenis syndrome (SMS) is a complex congenital anomaly characterized by craniofacial anomalies, neurological and behavioral disorders. SMS is caused by a deletion in region 17p11.2, which includes the RAI1 gene (90% of cases), or by point mutation in the RAI1 gene (10% of cases). Laboratory diagnosis is through cytogenetic analysis by GTG banding and molecular cytogenetic analysis by FISH. We carried out an active search for patients in Associations of Parents and Friends of Exceptional Children (APAE) of São Paulo and genetic centers in Brazil. Forty-eight patients were screened for mental retardation, craniofacial abnormalities and stereotyped behavior with a diagnosis of SMS. In seven of them, chromosome banding at high resolution demonstrated chromosome 17p11.2 deletions, confirmed by FISH. We also made a meta-analysis of 165 cases reported between 1982 and 2010 to compare with the clinical data of our sample. We demonstrated differences between the frequencies of clinical signs among the cases reported and seven Brazilian cases of this study, such as dental anom-
lies, strabismus, ear infections, deep hoarse voice, hearing loss, and cardiac defects. Although the gold standard for diagnosis of SMS is FISH, we found that the GTG banding technique developed to evaluate chromosome 17 can be used for the SMS diagnosis in areas where the FISH technique is not available.

**Key words:** Smith-Magenis syndrome; 17p11.2; FISH; Deletion