CNS and cutaneous involvement in tuberous sclerosis complex

Comprometimento cutâneo e do SNC no complexo esclerose tuberosa

Fernanda Frotte Bopp Salomon1, Miriam Menna Barreto1, Gláucia Zanetti1, Rosana Souza Rodrigues1,2, Emerson Leandro Gasparetto1, Edson Marchiori1

A 13-year-old boy with tuberous sclerosis complex (TSC) presented with a painless, gradually progressive area of thickened skin in the occipito-cervical region corresponding to a giant shagreen patch (Figure A). Physical examination demonstrates facial angiofibromas (Figure B) and periangual fibromas. He also presented mental retardation and tonic-clonic seizures. MRI revealed a subependymal nodule in the lateral ventricle, cortical tubers and the shagreen patch (Figures C and D).

The diagnosis of TSC is based on clinical criteria1,2. In our patient brain MRI was able to demonstrate at least three TSC major features, which met the criteria for a definitive diagnosis.

Figure. (A) A large shagreen patch located in the occipito-cervical region. (B) Multiple facial angiofibromas over the centrofacial areas. (C) Sagital T1-weighted and (D) Flair MRI shows subependymal nodules in the lateral ventricle close to the Monro foramina, suggesting giant cell subependymal astrocytoma at the right (arrows) and cortical tubers (arrowheads). Note also in C the shagreen patch (arrowheads).

References