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Clinical Neurology and Neurosurgery 108 (2005) 56–59

Clinical Neurology and Neurosurgery

www.elsevier.com/locate/clineuro

Case report

Intractable epilepsy in Turner syndrome associated with bilateral perisylvian hypoplasia: one case report

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Received 14 July 2004; received in revised form 22 October 2004; accepted 6 November 2004

Abstract

Turner syndrome (TS) is the most frequent sex abnormality in females, generally associated with a 45,X0 karyotype. Although neurological complications are frequently part of the clinical picture, serious brain abnormalities are quite rare in TS.

Epilepsy in TS is not frequent and so far only few cases have been reported, usually associated with cortical dysplasias. We report a Turner patient showing severe neurological impairment, refractory epilepsy and MRI finding of bilateral perisylvian hypoplasia. The possible dysgenetic role of X-chromosome on cortical morphogenesis is also discussed.

Keywords: Turner syndrome; Epilepsy; Cerebral dysplasia; X-chromosome; MRI

1. Introduction

Turner syndrome (TS) is a relatively common genetic disorder arising from X-monosomy or mosaicism. Physical features include short stature, webbing of the neck and gonadal dysgenesis [1]. Neurological complications are frequently found as part of the clinical picture, usually consisting of mental retardation, psychosis and neuropsychological dysfunctions [1–3]. Epilepsy is unusual in TS and so far only few cases have been reported, frequently associated with malformations of cortical development (MCD) [4–11]. Here, we present a Turner patient showing intractable epilepsy, severe cognitive impairment and congenital suprabulbar paralysis. A similar clinical picture has been recognized in the Literature related to a cerebral malformation known as congenital bilateral perisylvian syndrome (CBPS), and a bilateral perisylvian polimycrogiria is observed at MRI study. In our observation, MRI suggests a severe bilateral perisylvian hypoplasia.

2. Case report

This 22-year-old woman was adopted at age of 6 months. There was no history of neurological illness in her natural family, and she was born with normal labour at 38 weeks gestation. Severe developmental delay become soon evident; her parents report poor suck, excessive drooling and difficult in swallowing with recurrent respiratory infections. Motor milestones were severely delayed and she had poorly developed language skills. Later, progressive contractures of the joints, in particular of the knees, appeared, contributing to her motor handicap. Epilepsy began at about 1 year of life, with both generalized and partial seizures. These latter were characterized by gasps, facial expression of fear, vocalizations, right or left deviation of the head and eyes, followed by trashing hyperkinetic activity. Seizures occurred almost daily, more often during the sleep. Despite treatment with several anti-epileptic drugs and a 6 months trial with ketogenic diet, seizure control was never achieved. At the age of 18, the girl experienced a recurrent status epilepticus leading her to our observation. At general examination, she presented short stature, webbed neck, micrognathia, pterigium colli and multiple joint contractures. Mental retardation was severe but IQ could not

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doi:10.1016/j.clineuro.2004.11.012

be accurately assessed. Neurological examination revealed spastic quadriparesis, poor language skills and pseudobulbar signs, such as severe dysarthria, drooling and dysphagia; tongue movements were consistently restricted, with voluntary/automatic dissociation. Therapy with valproate and oxcarbazepine only reduced seizure severity.

3. Investigations

Cytogenetic analysis revealed a 45,X0 karyotype without mosaicism. Radiographs showed diffuse bone demineralisation and multiple arthicular deformities, with ankylosis in flexion of the knees. Bone maturation was significantly delayed, corresponding to the age of 15 years and 7 months. Interictal EEG showed diffuse slowing of background activity and sharp waves or sharp-wave and slow-wave complexes on the left fronto-temporal regions (Fig. 1A–C). Video-EEG monitoring recorded several, stereotyped, seizures: sudden change of respiratory rhythm and gasp, expression of fear, followed by tonic stiffening and elevation of the left arm. Seizure onset was accompanied by diffuse EEG flattening, followed by low-voltage recruiting rhythm more evident on the midline.

MRI was performed on GE Vectra 0.5 T with sequences SE DP/T2 weighted axial, IR T1 axial weighted and GE T1 axial, coronal and sagittal. It showed bilaterally enlarged ("wide-open") sylvian fissures with hypoplasic gyri of insular cortex. The underlying white matter appeared reduced in the volume but with normal signal intensity and digitations (Fig. 2A and B).

4. Discussion

TS is the most frequent sex abnormality in females, affecting approximately 0.2 per 1000 women [1]. It is generally associated with a 45,X0 karyotype, even if more than half patients have a mosaic chromosomal component (e.g., 45,X0/46,XX). Neurological symptoms are frequently part of the clinical picture: patients may show mental retardation, psychosis and specific neuropsychological dysfunctions,

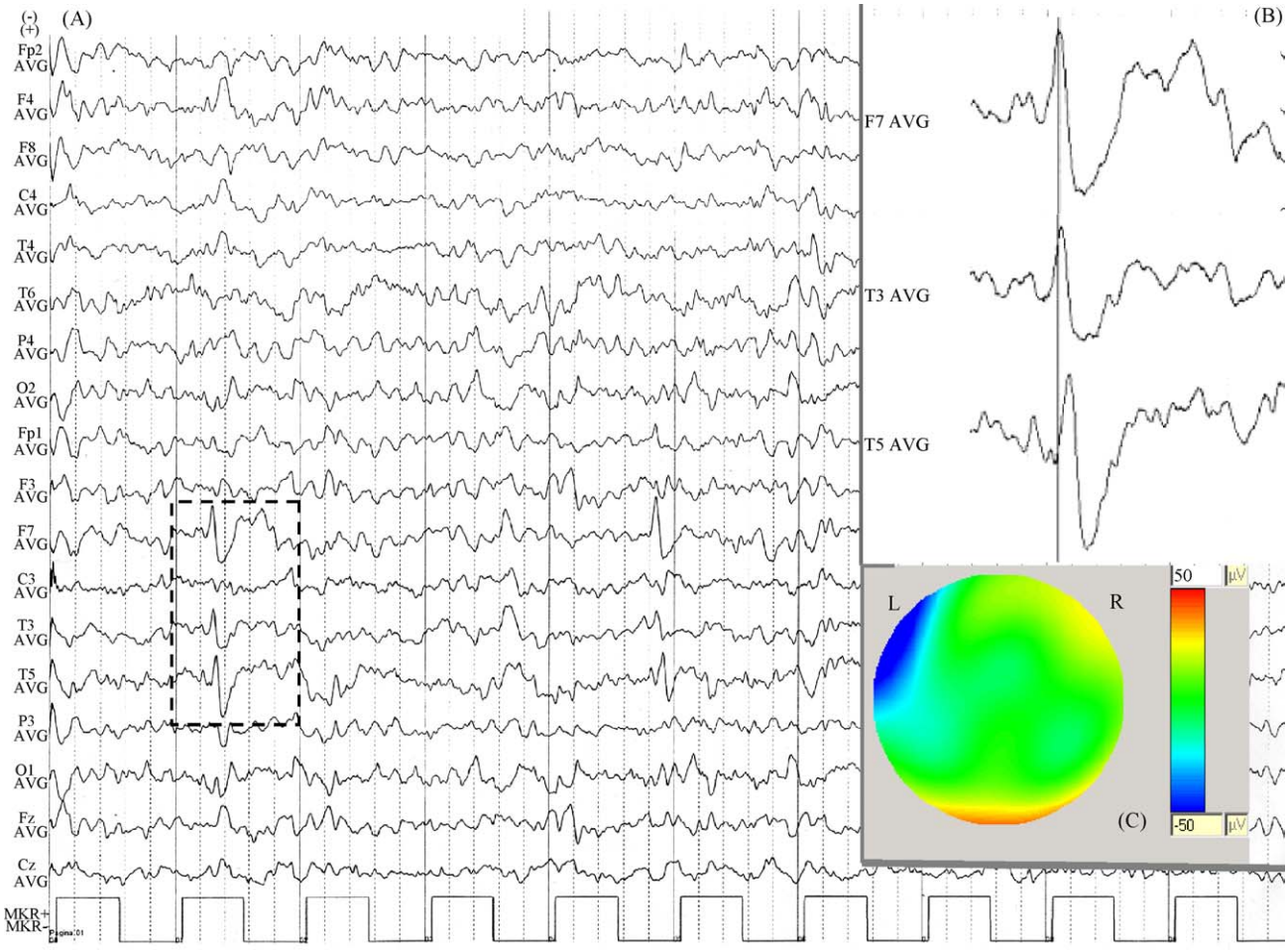

Fig. 1. Interictal waking electroencephalografic (EEG) recording shows diffuse slowing of background activity (in the theta range) and intermittent sharp- and slow-wave complexes on the left fronto-temporal areas (A). In the inset, the progression of the paroxysmal activity from the frontal (F7) to the medio-temporal (T3) and to the temporal posterior (T5) electrodes is shown (B). Amplitude map of paroxysmal activity (C).