This young boy is the first child of healthy non-consanguineous, white British parents.
He was born at term following an uneventful pregnancy weighing 3132 g (9th–25th centile).
Early development was thought to be normal.
At approximately 6 months of age he developed tonic upward eye deviation associated with flexion of his arms and neck consistent with infantile spasms.
An electroencephalograph (EEG) at this time was grossly abnormal (hypsarrhythmia) and strongly supported a diagnosis of West Syndrome.
Cranial MRI was reported as normal.
Prednisolone was prescribed and treated the seizures effectively.
Steroids were weaned over 6 weeks and he remained seizure free for a further 6 months.
By the age of 1 year, it was apparent that his early developmental progress was not being maintained and that he was functioning at the 6–8 month developmental stage.
Seizures returned shortly after his first birthday and were prolonged, frequent and on occasion focal, involving his right arm, leg and right side of face.
Clonazepam briefly improved seizure frequency, but subsequently his epilepsy has proved refractory to various combinations of anticonvulsant therapy.
Prolonged seizures of more than 60 min have been associated with a stepwise regression in his neurodevelopment.
Seizure semiology is now predominantly one of epilepsia partialis continua involving the right side of his face, right arm and right leg.
The development of focal seizures and the progressive nature of the condition prompted a second cranial MRI at the age of 2 years 6 months.
By contrast with the previous scan, this MRI revealed symmetrical subcortical white matter lesions (Fig.1A) with thinning of the anterior and genu of the corpus callosum (Fig.1B).
On examination, the patient had small, round, anteriorly rotated ears and a broad nasal root.
He demonstrated no visual awareness but startled to loud noise.
Tone was increased in all 4 limbs with internal rotation of both legs at the hips.
Reflexes were pathologically brisk.
Brief myoclonic jerks were evident throughout the examination.