Computed tomography in Sjögren-Larsson syndrome

J.M. Gomori1, V. Leibovici2, A. Zlotogorski2, I. Wirguin3, and S. Haham-Zadeh2

Departments of 1 Radiology, 2 Dermatology and 3 Neurology, Hadassah University Hospital, Jerusalem, Israel

Summary. A family with 12 children, 6 of whom had Sjögren-Larsson syndrome, was studied clinically. On CT, all six siblings with the disease had patchy regions of white matter hypodensity, most marked in the frontal lobes, which were confluent in the more severe cases. The ventricles were not enlarged and no other abnormality was demonstrated. The CTs of the 6 unaffected siblings and the mother, who was also unaffected by the disease, were normal.

Key words: Sjögren-Larsson syndrome - Cerebral computed tomography

Sjögren-Larsson syndrome is a rare syndrome with an autosomal recessive mode of inheritance, whose underlying biochemical defect has not yet been elucidated. About 200 cases have been reported, frequently from areas where the population is inbred, e.g., Vasterbotten County in Sweden and the Halifaxes of Halifax and Warren Counties in North Carolina, USA. It is characterized by congenital ichthyosis, spastic di- or tetraplegia, mental retardation, and difficulty with speech. It may be associated with anomalies of the eye, including a pathognomonic circular pattern of glistening white dots in the macular region of the retina, kyphosis secondary to muscular weakness, small stature, dental anomalies, and seizures [1-6]. The only previous computed tomography (CT) study of Sjögren-Larsson syndrome, involving five patients, was reported as yielding normal CT appearances [7]. This report presents our experience with cerebral CT in 6 siblings with Sjögren-Larsson syndrome.

Material

Of a family of 12 children, 6 had classical findings of Sjögren-Larsson syndrome. None had seizures. The parents were unaffected and were first cousins once removed from a village in a rural area near Jerusalem where there is a great deal of inbreeding (Fig. 1). All the children and the mother were studied clinically and by CT. The ages of the affected siblings were 25, 23, 22, 13, 8, and 5, and the ages of the unaffected siblings, 20, 14, 11, 10, 9, and 6 years. The most severe neurological findings were recorded in the 22-year-old. The 25- and 23-year-old siblings were equally and severely affected, and the 13-, 8-, and 5-year-old siblings were less markedly affected, the severity correlating with their ages. Unenhanced CT was performed in all 12 siblings and the mother with an Elscint 2002 (340 x 340 matrix, 10-mm-thick slice, 140 kVp, 8-s scan). The sibling with the worst neurological findings (the 22-year-old) also had a double-dose (80 g iodine) enhanced CT with the Elscint 2002 and another unenhanced CT on an EMI 1010 (160 x 160 matrix, 13-mm-thick slice, 140 kVp, 60-s scan time).

Results

The CTs of the affected siblings showed regions of white matter hypodensity that varied from frontal patches (Fig. 2) to a diffuse confluent involvement (Fig. 3). There was no associated calcium deposi-
Fig. 2a, b. CT of the 8-year-old Sjögren-Larsson syndrome patient demonstrates patchy foci of white matter hypodensity, which is most prominent in the frontal regions. c, d CTs of the affected 23-year-old sibling, showing similar findings.

Fig. 3a–c. CT of the neurologically most several affected sibling, 22 years old, demonstrating diffuse confluent white matter hypodensity.

Fig. 4a–c. Normal CT of the unaffected 20-year-old sibling.

tion. The severity of the CT findings correlated well with the severity of the neurological findings. The normal siblings and mother had normal CTs (Fig. 4). The sibling with the most severe neurological findings underwent a double-dose enhanced study which did not show any abnormal enhancement, and unenhanced study of this patient with the EMI 1010 showed a confluent white matter hypodensity similar to that seen on the Elscint 2002.

Discussion

Clinically, Sjögren-Larsson syndrome is readily diagnosed, because the clinical findings are pathognomonic and are associated with negative laboratory test results [4]. It must be differentiated on clinical grounds from the very rare Rud syndrome, which also involves ichthyosis and mental retardation but differs from Sjögren-Larsson syndrome in that there is polyneuritis and no spastic diplegia. It also was retinitis pigmentosa, seizures and frequently hypogonadism or eunuchism. Other syndromes with dermatological and neurological findings are: Refsum syndrome (ichthyosis, polyneuritis, cerebellar ataxia sometimes with paresis of the distal extremities, nystagmus, atypical retinitis pigmentosa, inner ear deafness, increased protein in the cerebrospinal fluid and evidence of 3,7,11,15-tetramethylhexadecanonic acid in the serum); maple syrup urine disease (quickly progressing mental deficiency, rigidity, spastic disorders, decerebration, pellagra-like photodermatosis, pallor of the optic discs, myoclonia, fainting fits and increased valine, leucine and isoleucine levels in the blood and increased keto-acid derivatives of these amino acids in the urine, usually culminating in death within the first few