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Neuroradiologic Findings in Marinesco-Sjögren Syndrome

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PURPOSE: **Our purpose was to determine the neuroradiologic findings of Marinesco-Sjögren syndrome on plain skull radiographs, CT, and MR images.**

METHODS: **Eight patients with proved Marinesco-Sjögren syndrome (age range, 4 to 56 years) had a total of nine CT scans, seven MR imaging studies, and two plain radiographic examinations of the skull. The findings were reviewed retrospectively, with particular attention to the size of the posterior fossa and cerebellum.**

RESULTS: **All patients had hypoplastic cerebellar hemispheres and a hypoplastic vermis in a small posterior fossa. One patient had a midline posterior fossa cyst and another had agenesis of the corpus callosum.**

CONCLUSION: **Hypoplasia of the cerebellar hemispheres and the vermis and a small posterior fossa are the most prominent neuroradiologic findings in Marinesco-Sjögren syndrome.**

Marinesco-Sjögren syndrome is a rare autosomal recessive disorder characterized clinically by cerebellar ataxia, congenital cataracts, mental and physical retardation, skeletal anomalies, and myopathy (1, 2). The recently described radiologic skeletal manifestations of the syndrome include kyphoscoliosis, club foot, short metatarsals and metacarpals, pectus deformities, gracile bones, microcephaly, and decreased bone age (1). We report the neuroradiologic manifestations of this syndrome on plain radiographs, computed tomographic (CT) scans, and magnetic resonance (MR) images in eight patients.

Methods

Our patient population consisted of 17 patients. Eight of these, five male and three female, 4 to 56 years old (mean age, 25 years), had imaging studies and were included in this study. All patients were treated at our institution over a period of 14 years, and all had mental retardation, cerebellar ataxia, and myopathy, as assessed by a clinical geneticist and reported previously (3). Four patients underwent muscle biopsies. Nine CT studies, seven MR examinations, and two lateral skull radiographs were performed, and were reviewed retrospectively. The Table shows the distribution of diagnostic radiologic procedures among the eight patients. Transaxial CT scans were obtained with 4- to 5-mm-thick sections through the posterior fossa and with 10-mm-thick sections through the cerebral hemispheres (window = 100, level = 50). Transaxial T1- and dual-echo T2-weighted MR images in 3- to 5-mm-thick sections and sagittal and coronal T1-weighted images in 6- to 8-mm-thick sections were obtained using a O.5-T or 1.5-T Picker MR unit. Sizes of the posterior fossa, cerebellar vermis, and cerebellar hemispheres were appraised as either normal or smaller than normal.

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Results

Clinically, all patients had mental retardation, cerebellar dysfunction, myopathy, cataracts, and dyslalia (slurred speech). As determined by the various imaging studies, all patients had hypoplasia of both the vermis and cerebellar hemispheres and a small posterior fossa. Patient 1 had platybasia (155° by Welcher's Basal Angle) and patient 2 had a posterior fossa cyst and hemorrhage of the left basal ganglia associated with prior trauma. Agenesis of the corpus callosum with colpocephaly in patient 7 probably was coincidental to the posterior fossa changes. Technical

Distribution of imaging studies among eight patients with Marinesco-Sjögren syndrome

Patient	Age, y/Sex	No. of Imaging Examinations		
		Skull Radiograph	MR	CT
1	23/F	1	2	2
2	33/F	...	1	1
3	4/M	...	...	2
4	56/M	...	2	1
5	8/M	...	...	1
6	29/M	...	1	...
7	20/M	1	1	1
8	23/F	...	...	1

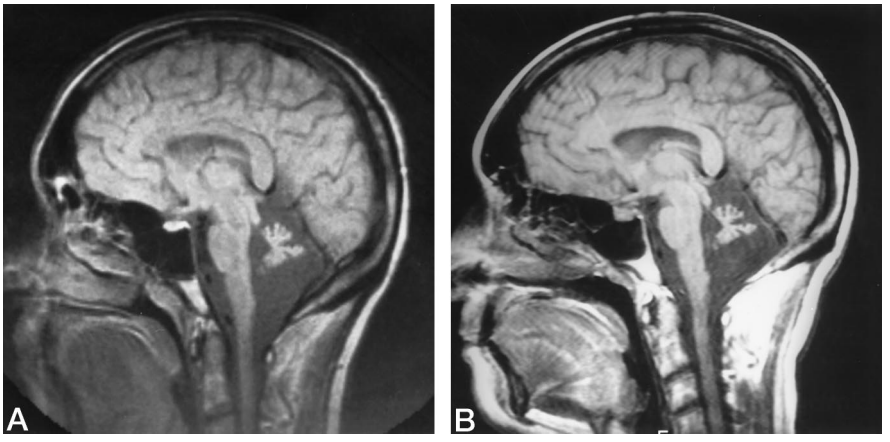

FIG 1. MR findings in a 56-year-old man with Marinesco-Sjögren syndrome (case 4).

A, Midline sagittal proton density–weighted image (789/40/1 [repetition time/echo time/excitations]) shows a small posterior fossa and hypoplastic cerebellum.

B, Midline sagittal T1-weighted image (600/20/1) 7 years later shows the same findings with no significant change.

variations in the different examinations over the years precluded performing quantitative measurements of the posterior fossa structures to examine the possibility of progressive atrophy over time. Subjectively, however, there were no significant changes in those patients in whom multiple studies were available for comparison over a span of 7 years (Fig 1).

Discussion

In 1931, Marinesco and coauthors (4) described four affected siblings in a Romanian family. Sjögren, in 1950, described the same syndrome in 14 Scandinavians, most of whom were offspring of consanguineous parents (5). Franceschelli et al (6), in 1956, presented a similar case and proposed labeling the syndrome with the names of the two senior authors of the earlier reports. Subsequent authors noted that the syndrome had been described in the Hungarian language more than 25 years before the appearance of Marinesco's article (2).

Marinesco-Sjögren syndrome is inherited as an autosomal recessive disorder, with most cases arising from consanguineous parents, although sporadic cases do occur. About 100 cases of Marinesco-Sjögren syndrome have been reported worldwide, mostly in the neurologic and genetic literature. All our patients came from an isolated rural area of southwest Alabama and were of Caucasian, African American, or Native American bloodlines, with extensive consanguinity; indeed, most could be traced to one of three family names (3). There are no specific genetic tests for Marinesco-Sjögren syndrome. Efforts to identify the genetic locus are ongoing.

The neuroradiologic hallmarks of the disease are cerebellar hypoplasia, a small posterior fossa, and absence of the lens subsequent to early cataract extraction. These findings should be differentiated from other causes of a small cerebellum with a normal posterior fossa, such as chemical toxicosis (alcohol, phenytoin, mercury) (7, 8); hereditary and degenerative diseases (olivopontocerebellar degeneration, hereditary ataxia, and ataxia telangiectasia) (7, 9, 10); vertebrobasilar insufficiency; paraneoplastic syndromes; and radiation to the posterior fossa (7, 11). Other causes of cerebellar dysfunction include infantile autism and fragile X syndrome (12, 13). In all these conditions, the cerebellum is small with a normal-sized posterior fossa, in contrast to Arnold-Chiari malformation, in which the normal cerebellum is compressed in a small posterior fossa. Marinesco-Sjögren syndrome may be differentiated from Arnold-Chiari syndrome by the presence of characteristic bony abnormalities, myopathy, and congenital cataracts. Other causes of congenital cataracts and mental retardation include myotonic muscular dystrophy (autosomal dominant) (14), ectodermal dysplasia (15), and chondrodysplasia punctata (15).

Conclusion

Marinesco-Sjögren syndrome (4) is a rare autosomal recessive disorder characterized by cerebellar ataxia, congenital cataracts, mental retardation, myopathy, and characteristic skeletal and neuroradiologic findings. The role of imaging is to suggest this syndrome in the differential diagnosis of a small posterior fossa or hypoplastic cerebellum and congenital cataracts. The presence of characteristic clinical findings and a history of consanguineous marriage help determine the final diagnosis.

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