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HEREDITARY SPINO-CEREBELLAR ATAXIA, OLIGOPHRENIA AND CONGENITAL CATARACTS

Report of two Cases of Marinesco-Sjögren Syndrome

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The association of spino-cerebellar ataxia, retarded mental and physical development, and congenital cataracts was first described in 1931 by Marinesco et al. in four Rumanian siblings; one boy and three girls in a family of six. Sjögren (1950) in Sweden was able to locate 14 affected individuals in six families. Ten were dead and he was able to examine the other four. His genetic analysis indicated that this is a hereditary disorder, transmitted by a single autosomal recessive gene and frequently resulting from a consanguineous marriage. Other cases with this condition have been reported by Richards, Garland and Moorhouse, Franceschetti et al, MacGillivory, Dogulu and Mutlu, Dureaux et al., Amyot, Géraud et al. (accepting only 2 of his 11 cases), and here in the United States by Alter et al., a total of 38 reported cases.

The purpose of this report is to describe two siblings with this syndrome. To the best of our knowledge this is the second reported family in the United States.

Figure 1 shows the pedigree of the two patients described below (VI 1 and 4). The propositus' family had 1 affected female, 1 affected male and 2 normal males (VI 2 and 3). There were 2 consanguineous marriages in two generations (IV and V). The grand-parents (IV 4 and 5) of the two patients on the mother's side were first cousins and their parents (V 1 and 2) were third cousins. The patients grandparents (first cousins) had 5 children (V2-6), 3 females and 2 males, and all were normal. V 2 was the patient's mother. Her other 4 siblings had a total of 34 descendants in two generations (VI and VII). Of these, 6 had congenital maldevelopments of the limbs, affecting one hand in VI 6, 12 and VII 22, both hands in VI 9 and VII 19, and both legs in VII 6. Two others, VII 3 and 4, had diabetes. This pedigree was constructed with the help of the patient's father and one brother.

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CASE REPORTS

CASE NO. 1 — This 42-year-old white male (Figure 2), the youngest of four siblings, was born at full-term after an uneventful delivery. He had no serious illnesses in the neonatal or infancy periods. Retardation in walking was noted, as compared with his normal siblings. As early as five years of age pes cavus, hammer toes and unsteadiness of gait were also noted. His speech was never normal and was described as explosive and slurred though he was able to communicate and apparently went to high school but did not graduate. From early childhood he had visual difficulties, and at age 13 was diagnosed as having cataracts in both eyes; these were eventually removed 17 years later. His clumsiness and weakness of the lower limbs as well as his speech difficulties became slowly and progressively worse through the years.

On examination, B. P., was 120 lbs and pulse 80 and regular. His height was 5 ft. 4 in. and weight 113 pounds. He was alert and oriented with dysarthric speech of an ataxic cerebellar type with bursts of intermittent clarity. His voice was hoarse and the general tone of speech was hypotonic. Wechsler Adult Intelligence Scale revealed a verbal I.Q. of 54 and a performance I.Q. of 50. The fundi were not visualized. Nystagmoid movements were present on lateral gaze. He was almost unable to stand without support and there was marked ataxia of gait. Limb ataxia was mild in the lower extremities and not obvious on finger-to-nose test though mild dysdiadochokinesis in the arms was present. He had bilateral mild foot drop with very high pes-cavus and hammer toes. The lower limbs were hypotonic yet with hyperactive knee jerks, abortive knee clonus, hypoactive ankle jerks, and bilateral Babinski and Chaddock signs. The stretch reflexes were normal in the upper limbs. Position sense was absent in both feet, and vibration sense was absent up to the lower dorsal spine. The spino showed dorsal scoliosis and lumbar lordosis. Laboratory studies included urinalysis, white count, differential count, hemoglobin, blood urea nitrogen level, calcium, phosphate, 2 hs. pc blood sugar and peripheral blood smear, all of which were normal. Protein electrophoresis: total protein 6.9, albumin 2.83, alpha-1 globulin 0.26, alpha-2 globulin 0.84, beta globulin 1.17 and gamma globulin 1.80 Gm/100 ml. The E.K.G. indicated left ventricular hypertrophy. The skull X-rays were normal, and chest X-rays were negative except for moderate dorsal scoliosis with convexity to the right. The E.E.G. showed generalized disturbance of cerebral rhythms without any localizing or paroxysmal features.

CASE NO. 2 — This 58-year-old white female (she was not examined personally by us), the eldest of four siblings was born at full term. As early as six or eight years of age she was known to have pes cavus, unsteadiness of gait and “explosive speech”. She did not go to school because of visual deficit and at age 10 was diagnosed as having cataracts in both eyes which were subsequently removed. She was described by her family as “never being very bright”. Her clumsiness, ataxia and dysarthric speech have slowly but progressively worsened. She was married but never had any children and her mother and relatives have taken care of her and her duties for most of her life.
Figure 2

Photographs of patient reported as case 1.
Review of the literature revealed 38 reported cases of Marinesco-Sjögren syndrome characterized by spino-cerebellar ataxia, retarded mental and physical development and congenital cataracts. Of the 40 affected cases (including ours), 18 were males and 22 females. Of 19 families with affected children, 12 had consanguineous parents, most often first cousins. Cerebellar ataxia has been the first recognizable feature of the disorder, mainly ataxia of gait and stance. Most of the cases have shown nystagmus and different degrees of cerebellar dysarthria. Bilateral cataracts frequently detected in early childhood, and moderate to severe mental retardation without gross evidence of progressive deterioration were always present. In one of our cases there is a suggestion of progressive mental retardation, since at age 42 his I.Q. was 50 and allegedly he was able to pass through high school in his teens. All the patients were also undersized. Skeletal abnormalities include kyphoscoliosis, genu valgum, deformities of digits, pes cavus, pes planovalgus, talipes equinovarus and pes planus. Weakness affecting mostly the lower limbs and cortico-spinal tract involvement with hyperactive and brisk stretch reflexes and Babinski signs have also been reported. A post-mortem examination has not yet been reported; the only pathological material available is a biopsy carried out on the right frontal cortex of one of Marinesco's patients, which showed atrophy of ganglion cells and a paucity of nerve fibers. There is nothing to suggest that this syndrome necessarily shortens the life span.

Further reports and more definite pathological evidence will be necessary for a better understanding of this syndrome and its relationship with the group of spino-cerebellar degenerative disorders. We feel, however, that the association of spino-cerebellar ataxia, retarded mental and physical development, congenital cataracts and the high incidence of consanguineous ancestors suffice to differentiate this syndrome clinically from other disorders of the spino-cerebellar degenerative group.

SUMMARY

Two siblings with spino-cerebellar ataxia, retarded mental and physical development congenital cataracts and a family history of consanguineous marriages are reported.

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SPINO-CEREBELLAR ATAXIA

REFERENCES


