

letters

A syndrome of congenital ichthyosis, mental retardation, myopathy and anemia in dizygotic twin sisters

To the Editor: In 1927, Rud¹ described a patient with congenital ichthyosis, hypogonadism, short stature, epilepsy, polyneuritis, and hyperchromic macrocytic anemia. In the following years, 54 patients in 36 published reports were reported as having Rud syndrome.²⁻⁵ Recently it has been argued that the various case reports on Rud syndrome were so genetically heterogeneous and differed greatly from the original case reports of Rud.⁶ We present a report on dizygotic twin sisters with congenital ichthyosis, mental retardation, myopathy and hyperchromic macrocytic anemia.

The patients were 21-year-old female and dizygotic twins. Their parents are relatives of the first degree. The mother had three pregnancies. The first one ended with abortus and the second one with a healthy birth. The births were term after an uneventful pregnancy and normal delivery with birth weights of 3300 g and 3200 g. They were affected with ichthyosis from birth. The twins' psychomotor development was delayed. They managed to walk at the age of 4 with persisting difficulty in running and climbing stairs. They graduated from a high school for the mentally handicapped and had mild psychomotor retardation (IQ=69). The family history includes an aunt with progressive myopathy and mental retardation who had died at the age of 38 years.

The examination of the skin showed signs of generalized moderate erythema and fine scaling, involving the scalp, face, trunk and extremities (**Figure 1**). Our cases had thickened and hyperkeratotic palms



Figure 1. Ichthyosis of the belly skin.

and soles and mild alopecia over the scalp. Even though they had dry eyes, the other ophthalmic and odyometric examinations showed no abnormal findings.

The twins had mild hyperchromic macrocytic anemia despite vitamin B12 injection therapy from the age of 11 years. Other laboratory findings including serum phytanic acid levels were normal. Electrocardiogram and electroencephalogram results were normal and myopathy was diagnosed by electromyography. In the cranial MRI, cerebellar vermicular folia were minimally dilated and deep. The fourth ventricle was in the midline, and lateral ventricles were asymmetric. Both of the structures were dilated (**Figure 2**).

Our cases had congenital ichthyosis, alopecia, mental retardation, anemia and myopathy. They would have been classified as Rud syndrome earlier, but recently the Rud syndrome diagnosis has been eliminated because of the heterogeneity of the new cases and differences from Rud's original description of the syndrome. The cases differ from Refsum disease by having normal



Figure 2. Cranial MRI of one of the twin sisters.

levels of phytanic acid, mental retardation, and myopathy that are not the characteristics of Refsum disease.⁷ Sjögren-Larsson syndrome was ruled out since they showed no sign of spastic quadriplegia, retinal dystrophy, phenotypically and electrophysiologically findings that are identical for this syndrome.⁸ Our cases also had little similarity to any of the other ichthyosis syndromes like Tay syndrome,⁹ Netherton syndrome,¹⁰ multiple sulfatase deficiency,¹¹ neutral lipid storage disease,¹² hereditary sensory neuropathy,¹³ Brocq syndrome,¹⁴ Zunic neuroectodermal syndrome,¹⁵ ichthyosis-mental retardation-dwarfism and

renal impairment,¹⁶ or cardiofacio-cutaneous syndrome.¹⁷

In conclusion, the two cases seem to be different from any previously recognized and currently known syndromes of dizygotic twin sisters.

Burcu Karaca

Biga Devlet Hastanesi FTR
Poliklinik, Canakkale
Kibris Sehitleri Caddesi Sirintepe
Mevkii, Biga Canakkale 17200
Turkey
drburcub@yahoo.com

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