

Encephalopathic Susac's syndrome associated with livedo racemosa in a young woman before the completion of family planning. **BMC Neurol** 2013 Nov 25;13:185).

COMMENTARY. Susac syndrome (SS)[1] consists of a triad of encephalopathy, branch retinal artery occlusions and hearing loss. Associated abnormalities include multifocal corpus callosal lesions on MRI [2], resembling a vasculitis, and autoimmune disorder such as juvenile dermatomyositis. Headache is usually constant but was absent in the above case. Women are affected more often than men (3:1); the age of onset ranges from 7 to 72 years, but ages 20-40 are most vulnerable [3]. SS is an autoimmune endotheliopathy that responds to treatment with immunosuppressants, steroids, cyclophosphamide, and IV immunoglobulin, with aspirin as an adjunct [2][4].

### References

1. Susac JO, et al. *Neurology*. 1979 Mar;29(3):313-6.
2. Susac JO, et al. *J Neurol Sci*. 2007 Jun 15;257(1-2):270-2.
3. Rennebohm R, et al. *J Neurol Sci*. 2010 Dec 15;299(1-2):86-91.
4. Millichap JG. *Neurological Syndromes*. New York: Springer; 2013:279.

## INTERFERON BIOMARKERS IN AICARDI-GOUTIERES SYNDROME

Investigators at University of Manchester, UK, and multiple international centers studied interferon-related biomarkers in patients with Aicardi-Goutieres syndrome (AGS). Of 82 patients with AGS, 74 (90%) had a positive interferon score. The measurement of an interferon score might be used to assess efficacy of anti-inflammatory therapy. (Rice GI, Forte GMA, Szykiewicz M, et al. Assessment of interferon-related biomarkers in Aicardi-Goutieres syndrome associated with mutations in TREX1 (and any of six genes): a case-control study. **Lancet Neurol** 2013 Dec;12(12):1159-69).

COMMENTARY. Aicardi-Goutieres syndrome is an early-onset familial encephalopathy characterized by brain atrophy, microcephaly, spasticity, dystonia, psychomotor retardation, chronic CSF lymphocytosis, basal ganglia calcification, autoimmune disorders such as chilblains (pernio), and increased interferon-alpha in the CSF and serum [1][2]. High mortality in the first year is common. An early active stage of the disease followed by a period of attenuation correlates with higher levels of interferon activity in infancy and lower levels with increasing age.

### References.

1. Aicardi J, Goutieres F. *Ann Neurol*. 1984 Jan;15(1):49-54.
2. Stephenson JB. *Eur J Paediatr Neurol*. 2008 Sep;12(5):355-8.