English summary

European Susac Consortium (EuSac)

In Susac syndrome (occasionally also referred to as SICRET syndrome or RED-M), presumably autoimmune mediated occlusions of microvessels lead to a characteristic clinical triad of central nervous system dysfunction, branch retinal artery occlusions (BRAO), and sensorineural hearing impairment. The disease is named after John O. Susac who was the first to describe this condition in 1979. Since then, more than 300 cases have been reported from all continents. The pathogenesis is not yet clear. Autoimmune processes leading to damage and inflammation-related occlusion of the microvessels in brain, retina, and inner ear are thought to play a causal role. Considered a rare but important differential diagnosis in numerous neurological, psychiatric, ophthalmological, and ear, nose and throat disorders, better understanding of Susac syndrome is crucial.

The diagnosis of Susac syndrome is straightforward, when the characteristic clinical triad is complete, when the physician is familiar with the clinical presentation, and when the crucial diagnostic procedures are carried out and show characteristic findings like BRAO in retinal fluorescence angiography. However, the diagnosis is often complicated by the fact that in the majority of cases the characteristic signs do not occur concomitantly but rather develop successively with symptom-free intervals, which often enough results in a delayed or even completely missed diagnosis. Consensus criteria for the diagnosis of Susac syndrome have not yet been established.

Due to some overlap in the clinical presentation and the patterns of MRI pathology multiple sclerosis (MS) is probably the most frequent misdiagnosis of Susac syndrome. However, with respect to the different therapeutic approach, particularly the necessity of a first-line immunosuppressive treatment in Susac syndrome in contrast to primarily immunomodulatory approaches in MS, a prompt establishment of the diagnosis is essential. Additional diagnostic criteria allowing an early differential diagnosis are therefore highly warranted.

In the context of these challenges a Germany-wide network of Susac syndrome researchers and clinical neurologists was initiated in 2011 in order to merge and facilitate clinical and research activities around Susac syndrome. To date, four German university-based centers are actively participating (Münster, Berlin, Düsseldorf, Heidelberg). As the recruitment of additional collaboration centers in Europe is planned, a more international name, European Susac Consortium (EuSac), was chosen for the network.

The main aims of EuSac are:

- Improving the publicity of Susac syndrome
- Providing a platform for expert advice to Susac syndrome patients and referring physicians
- Establishing a national and international network for research activities on Susac syndrome.
- Investigating the pathophysiology of Susac syndrome
- Improving the diagnosis of Susac syndrome, in particular establishing consensus criteria for the diagnosis.
- Long term follow up on patients with respect to prognosis and outcome of Susac syndrome
- Biobanking for scientific evaluation of diagnostic and prognostic markers