

Autoimmune encephalitis – a newly recognised clinical manifestation of autoimmune polyendocrine syndrome type 1?



J. Ferenczova, V. Vargova, D. Krysl¹, E. Banoova, E. Sadova

Department of Paediatrics, Faculty of Medicine, Safarik University, Kosice, Slovakia

¹ Department of Clinical neurophysiology, Sahlgrenska University Hospital, Gothenburg, Sweden



INTRODUCTION

Autoimmune polyendocrine syndrome (APS) type 1 is a rare disease. The classic features are chronic mucocutaneous candidiasis, hypoparathyroidism and adrenocortical failure, but many other autoimmune disease components occur less frequently. There is an enormous variation in presentation and phenotype, which makes the diagnosis difficult. Several non-classic presentations of the disease have been described over the last few years.

Autoimmune encephalitis (AE) denotes a heterogeneous group of immune-mediated CNS disorders with characteristic clinical symptoms including epileptic seizures, memory dysfunction and psychiatric problems. AE can be divided into: 1) syndromes with antibodies against intracellular antigens (e.g. GAD65+) and 2) syndromes with antibodies against membrane and synaptic antigens.¹

CASE REPORT

14 - years old girl with APS type 1 presenting with typical clinical symptoms (Fig.1) was admitted to hospital for prolonged seizures. Hypoglycaemia and hypocalcaemia were excluded as a cause of the seizures. EEG showed findings non-specific for epilepsy. Examination of the CSF was performed to exclude infection (herpetic viruses, etc.), which all were negative. We extended our diagnostic spectrum of tests by screening autoimmune etiology of the seizures. We investigated blood and CSF for paraneoplastic antibodies (anti- Hu, Ri, Yo, CV2, Amphiphysin, Titin) and anti-GAD antibodies. Antibodies against GAD were positive in CSF (>2000 IU/L, (ref.range 0-10)) as well as in blood (>2000 IU/L (ref.range 0-10)). MRI has shown in T2 and FLAIR sequence hyperintensity. MRI angiography did not show vasculitis or other organic changes of cerebral vessels. We started with immunosuppressive therapy (glucocorticoids and i.v. immunoglobulins) and antiepileptics. Therapy resulted in a clinical course without seizures. After 6 months lasting treatment we repeated CSF and blood examination – anti GAD antibodies decreased in CSF (17 IU/L), but stayed same in blood (>2000 IU/L). Control MRI in the same time showed completely resolved pathological findings (Fig.2).

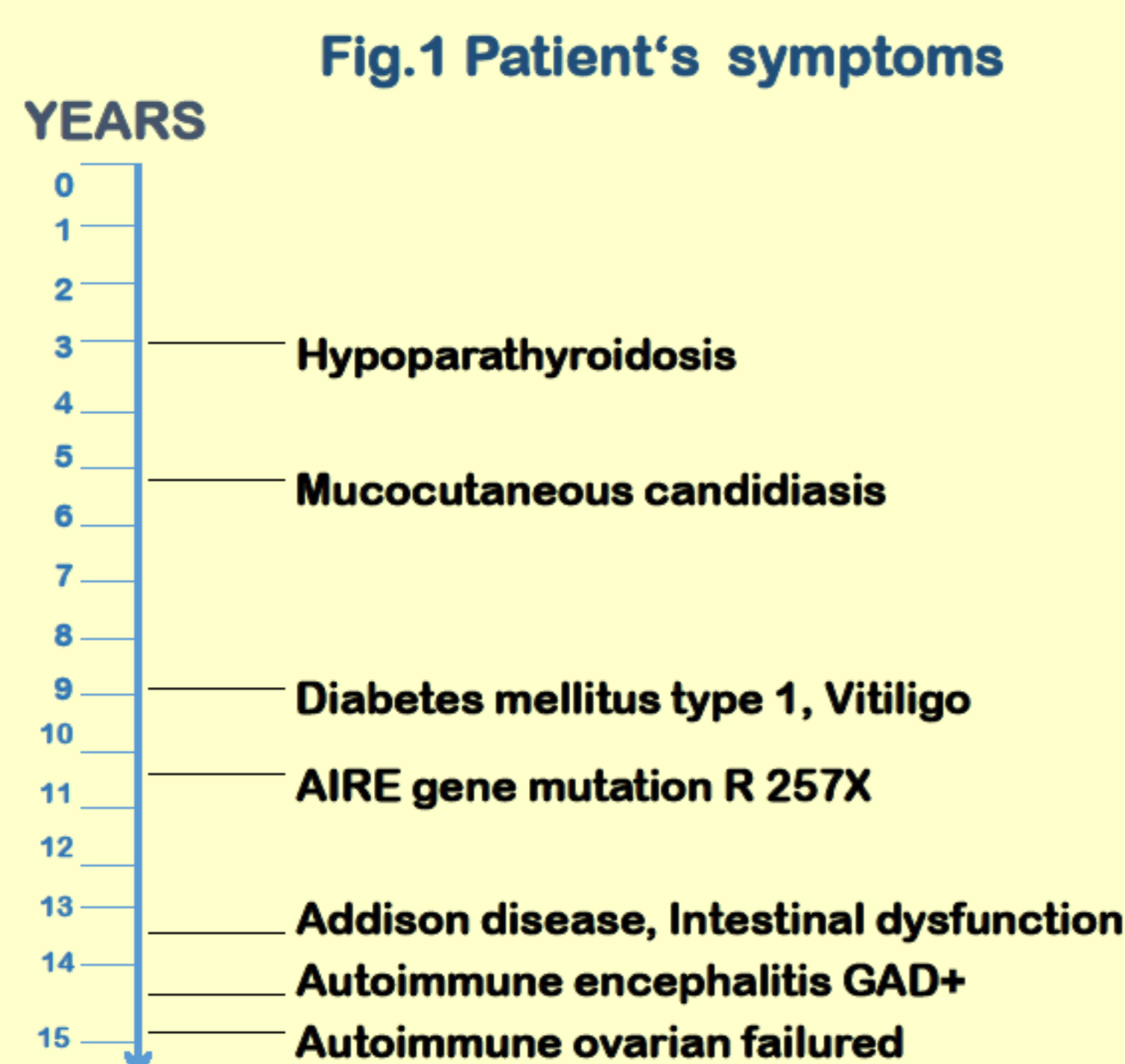
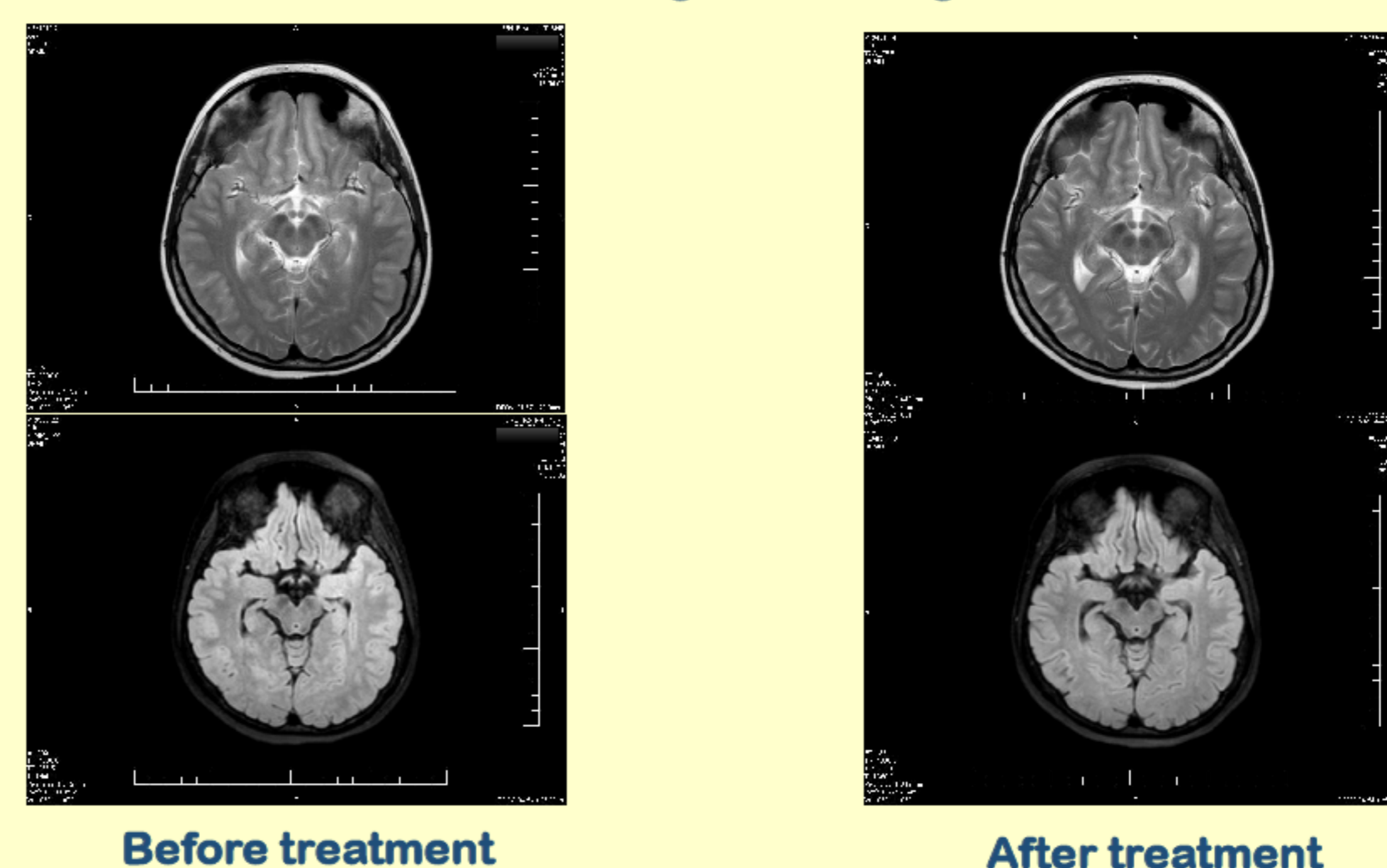


Fig.2 MRI imagine



DISCUSSION

Recently have been included among the APS 1 manifestations: chronic lung disease, chronic inflammatory demyelinating polyneuropathy and gastrointestinal dysfunction. For each of these novel components of APS 1, specific autoantibodies have been identified.² Autoimmune encephalitis is still underdiagnosed neurological disorder. Widening in the field of neuronal autoantibodies is a new diagnostic tool for early diagnosis and adequate therapy. At present, GAD65 positive AE was only a few times described as a part of other autoimmune disease/syndrome (e.g. diabetes mellitus type I).^{3,4}

CONCLUSION

This is first documented case of APS 1 in Slovakia. To the best of our knowledge this is the first described case of AE GAD65-positive as a component of APS 1. Early diagnosis and successful immunosuppressive therapy can lead to completely resolve. Autoimmune GAD65-positive encephalitis should be considered a new component of the clinical spectrum of APS 1.

REFERENCES

- 1 Krysl D et al: Autoimmune limbic encephalitis. *Neurol.praxi*, 2012;13:160-164.
- 2 Valenzise M et al.: APECED syndrome in childhood: clinical spectrum is enlarging. *Minerva Pediatr.* 2014 Dec 12.
- 3 Kuchling J, et al.: GAD-antibody-associated late-onset ataxia in two female siblings. *Cas Rep Neurol.* 2014;6:264-70.
- 4 Suleiman J et al.: Autoimmune epilepsy in children: case series and proposed guidelines for identification. *Epilepsia.*2013;54:1036-45.

