STIFF-PERSON SYNDROME WITH NEGATIVE ANTIBODIES: A CASE REPORT

Stefania Martina Angelocola*, C. Rinaldi*, M. Acciarri*, F. Girelli*, C. Cagnetti*, P. Di Bella*, L. Provinciali*, F. Logullo **

*Department of Experimental and Clinical Medicine - Marche Polytechnic University – Ancona **
*Department of Neurology - Provincial General Hospital – Macerata ***

INTRODUCTION

Stiff-person syndrome (SPS) is a rare autoimmune neurological disorder characterized by rigidity of axial and proximal appendicular muscles, abnormal gait and painful spasms.
It may be associated with **autoimmune disorders** such as insulin-dependent diabetes mellitus, Hashimoto's thyroiditis, pernicious anemia, and vitiligo; moreover, it can develop as a **paraneoplastic neurological disease**. It has been associated with a growing number of antibodies, such as: anti-GAD, anti-Glycine, anti-GABA-A or GABA-B, anti-Amphiphysin, anti-Ri, anti-Gephyrin.

CASE REPORT

-14-year-old male patient
-family members affected by amyotrophic lateral sclerosis (grandmother and maternal uncle)
-he gradually developed: difficulty in opening and closing his hands and a gait disturbance with reduced fluidity of his movements, associated with axial and proximal stiffness, cramps and legs pain. Morover his clinical history was also characterised by extrasystoles.
-**Electromyography** showed continuous motor unit activity and the co-contraction of agonist and antagonist muscles.
-**Creatinkinase** were more than 600 IU/L.
-**Brain MRI** was negative for pathological findings.
-**VGKC antibodies (LGI1 and CASPR2)** were assayed, in particular anti-CASPR2, in the Isaacs syndrome suspicion, with a negative result.
-**Anti-neural antibodies** (including anti-Amphiphysin and anti-Ri), anti-GAD, anti-pancreatic islet cell, anti-ANA, ENA, ANCA, anti-striated muscle were negative.
-**Genetic tests for ALS** resulted negative.

SPS was diagnosed resulting from clinical examination and electromyography, despite the antibodies tested negative, in particular anti-GAD65.
The patient was treated with benzodiazepines and baclofen with mild response; subsequently he started therapy with intravenous immunoglobulins and steroids, with improvement in his clinical condition.
The patient will have to undergo periodic cancer screening and cardiologic follow-up because of the frequent association of SPS with tumors and with autonomic disorders such as cardiac arrhythmia.

DISCUSSION AND CONCLUSIONS:

SPS is generally associated with specific antibodies, especially anti-GAD65; however we can also find cases without associated antibodies: this case report highlights the importance that clinical manifestations are essential for diagnosis of SPS.
Beside the immunology tests, electromyography is a very important diagnostic tool and reveals continuous motor unit activity that disappears during sleep and general anesthesia.

REFERENCES

1) Corina Roman Filip et al - "Stiff-Person Syndrome with negative anti-Gad65 antibodies: case report" - Romanian Journal of Neurology - 2013 - Vol XII N 3 - pag 156-159
2)Dalakas MC - "Stiff person syndrome: advances in pathogenesis and therapeutic interventions"- Current Treatment Options in Neurology – 2009 Mar - 11(2):102-10
3)Harini Sarva et al - "Clinical Spectrum of Stiff Person Syndrome: a Review of Recent Reports" - Tremor and Other Hyperkinetic Movements - 2016 - 6: 340

Sin

**XLVII CONGRESSO NAZIONALE
22-25 OTTOBRE 2016 – VENEZIA**