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Behind the weakness

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Background: 33 years old man with no allergies, renal insufficiency with nefrocalcinosis, congenital immunodeficiency (severe B white cellules deficit), type 1 Arnold-Chiari and ventricle peritoneal derivation, Stevens-Jhonsons syndrome and chronic lumbalgia. During the previous week the patient had been suffering from lumbar pain with increasing intensity despite the treatment. He started feeling weak and dizzy and even falling at home, so he had his derivation device checked and was transferred to his Hospital for observation. In the days that followed the weakness get worse, turning walk and stand up harder and showing urinary incontinence, disfagy and mental confusion.

Method: Physical exploration: Good overall status. Blood pressure 100/60 mmHg. Conscientious and orientated, no aphasia, facial asymmetry with right facial hypoesthesia, strength decreased in extremities, worsen in the more distal areas, with disproportional dissymmetry, dysestesia and abolished reflexes. Unstable sitting without ability to walk. No increased pressure at derivation device. Complementary tests: Blood test: urea 103, creatinin 2.11, C reactive protein 5.7, Leucocytes 12.320 (normal formula), IgA 183. Other determinations turned out normal. Cephalic raquideous liquid transparent with ADA 14, leucocytes 7, glucose 65, proteins 214, serological and immunological test negatives. Thorax abdominal CT and Cranial and cervical MR: no changes with previous. Electromiogram: proximal segments alterations in motor fibres conduction. Differential diagnosis: Infectious acute ventriculitis in derivation carrier, Meningeal infection, Derivation obstruction and hydrocefalia, inflammatory demyelinising acute pathology.

Results: Miller-Bickerstaff Syndrome. High range antibioterapy and endovenous immunoglobulins treatment were settled with a progressive remission of the motor symptoms and cognition. However a persistent facial paresis and slight sensibility alterations in feet get established.

Conclusions: Rare diseases diagnosis in chronic patients with multiple pathologies are often delayed due to confusing symptoms that could be related both to the acute and chronic process, leading to long term recoveries and sequels in patients.