ACUTE INTERMITTENT PORPHYRIA IN ELDERLY UNDERGOING HEMODIALYSIS: RESOLUTION OF TETRAPLEGIA WITH SYSTEMIC HEMIN AND REHABILITATION.

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INTRODUCTION: Porphyrias are inherited defects in the biosynthesis of heme. Attacks in Acute Intermittent Porphyria (AIP) are characterized by abdominal pain, neurological disturbances and psychiatric disorders and, in severe cases, they may lead to respiratory paralysis and coma. Porphyria has a mortality of 20-25% within the first five years since the first attack; rare in children and after the fifth decade. The reasons behind the attacks can be several: drugs, alcohol, stress, fasting, menstrual cycle, infections. The prevalence of acute porphyria is 10,1/100000 (according Orphanet, November 2016).
Renal involvement in acute porphyrias is represented by hyponatremia, urinary retention, tubulo-interstitial nephropathy, hypertension and chronic kidney disease. Most of our patients were affected by renal colic associated with pallor, nausea, vomiting, fever, acute retention of urine and dark urine.

A 68-year-old women, undergoing tri-weekly hemodialysis since January 2011 (32 months) for Chronic Kidney Disease (CKD) due to undiagnosed nephropathy has been diagnosed with Acute Intermittent Porphyria (AIP) after a family screening.
In October 2012 she underwent superior-external quadrantectomy of the right breast for invasive ductal carcinoma, grade II, score 6 (3, 2, 1) and intraductal papillomas (G2 pT2 pNOs).
In February 2013 she underwent subtotal thyroidectomy for goiter with focus of papillary microcarcinoma and subtotal parathyroidectomy for hyperplastic parathyroid.
In September 2013 she was admitted to the division of Nephrology because of abdominal pain, constipation and uncontrolled hypertension. Since she had no diuresis, plasma porphyrins were measured at a peak of 619 nm.
The patient reported depression and progressive muscle weakness in legs and then, the following day, even in arms, defining a medical case of flaccid tetraparesis. In suspect of poliradicoloneuritis, a lumbar puncture was made and it was negative. Two days later, hemin (Normosang) was administered through femoral vein catheter in order not to damage her arteriovenous fistula, at a dose of 3 mg/kg/24h for 4 straight days, and then bi-weekly for the following 2 months, during which she was moved to the division of Rehabilitation Medicine and Neuro-rehabilitation Unit where she started a rehabilitation plan since her limbs strength had a MRC-score of 0.

METHOD: Functional evaluation was assessed by Barthel scale (BS), at admission and at discharge. The BS quantifies global functional recovery and the degree of independence of any help (physical, verbal,…) in activities of daily living. It ranges from 0 to 100, with 0 indicating a totally dependent patient in bedridden state and 100 indicating that the patient is fully independent. Items are divided into groups that relate to self-care (feeding, grooming, bathing, dressing, bowel and bladder care and toilet use) and mobility (ambulation, transfers and stairs climbing). Muscular strength of involved paretic muscles was ascertained by use of the Medical Research Council (MRC) scale according to De Jonghe's method that, for each limb, evaluates muscular groups of three muscles: deltoid, biceps and wrist's extensor (upper limb); iliopsoas, quadriceps femoris and tibialis anterior (lower limb). The evaluation was performed at admission, 2, 4 weeks and at discharge. According to MRC scale, strength ranges from 0 (paralysis) to 5 (normal strength) for any muscle, so for each muscle group the score ranges from 0 and 15 0-5 for muscle bundle for 3 muscle groups for the 4 limbs respectively), making the overall score range from 0 to 60. A score less than 24 shows severe muscular deficit, whereas an MRC score with 48 or higher was considered a very mild weakness or normal. The subject underwent rehabilitation including joint mobilization, muscular stretching, proprioceptive neuromuscular facilitation, and occupational therapy. Furthermore, the patients had low frequency (4 Hz) electrical stimulation (ES) done, through COMPEX instrument, with the following physical parameters: 90 mA intensity and rectangular pulses of 0.2 ms were applied on quadriceps and tibialis anterior of lower limbs one hour daily, six day a week for one month. The patient underwent rehabilitation including electrical stimulation and occupational therapy

RESULTS: At admission, BS score was 25 indicating severe disability. Likewise, neurological picture showed severe strength impairment characterized by tetraplegia. MRC score was 0 either both upper and lower limbs. After the administration of hemin and rehabilitation treatment, muscular deficit progressively improved and MRC scores were 24, 36 and 50 at 2, 4 weeks and at discharge, respectively. Likewise, good functional outcome was also observed as BS scores were 45, 65 and 90 at 2, 4 weeks and at discharge, respectively. Equally, functional appearance is significantly improved as observed from the increase of BS scores to 45, 65 and 90, at 2, 4 weeks and at discharge, respectively compared at score of 25 at admission.

	2 weeks	4 weeks	discharge
BS	45	65	90
MRC*	24	36	50

BS	from 0 to 100	(0 = total dipendence – 100 = fully indipendence) (Barthel scale)
MRC*	from 0 to 60	(<48 = muscular deficit – > 48 = muscle weakness) (Medical Research Council)
		*cumulative value of upper and lower limb strength according to De Jonghe method

Three months after the beginning of the disease, she was given hemin for 4 straight days due to hyponatremia (132 mmol/l) and then monthly in addition to physical rehabilitation, until she improved her muscle strength and motor skills to ensure can go back to an autonomous life.

CONCLUSIONS: Currently, from a clinical and functional point of view, the patient presents: a) a good muscle tropism and tone, b) good articular function and c) self-walking capability with enlarged base at the slightest uncertainty. Right now the patient is given hemin every 2 months and we're planning to cut down even more the administration of hemin in the following months. The patient will keep on doing physical treatment and will follow a normocaloric, hyperglucidic diet with the addition of maltodestrins. Currently she is undergoing hemin therapy every 4 months and tri-weekly hemodialysis.

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