

Case Report

Malingering and Factitious Disorder (Münchausen-syndrome) can be Mitochondrial

Josef Finsterer, Stefan Lässer

ABSTRACT

Malingering and factitious disorder (Münchausen-syndrome) has not been reported as a manifestation of a mitochondrial disorder (MID). Here, we report a 46 years-old female with a MID due to a combined complex I-IV defect, manifesting in the cerebrum, muscle, bone marrow, kidneys, and the endocrine glands. Myopathy showed up as myalgia, easy fatigability, ptosis, and abnormal muscle biopsy. Endocrine involvement manifested as short stature and thyroid dysfunction. Involvement of the kidneys manifested as mild Fanconi syndrome. Bone marrow affection resulted in iron-deficient, chronic anemia with elevated transferrin. Cerebral manifestations included epilepsy with rare epileptic and frequent psychogenic seizures, and malingering and factitious disorder (Münchausen-syndrome). Cerebral magnetic resonance imaging was normal. Since malingering-disorder dominated the phenotype, the patient was in the majority of the cases not taken seriously, resulting in 175 hospital admissions over 20 years, repeated expensive diagnostic work-ups and huge amount of avoidable interventions. MIDs also manifest as malingering personality disorder (Münchausen-syndrome) but normal cerebral imaging. Management of such patients could be difficult for institutions not familiar with MIDs and facilitated and improved if these patients are taken seriously and treated at departments particularly dedicated to handle MIDs.

Key words: *Epilepsy, lactate, metabolic myopathy, mitochondrial disorder, multi-organ disorder syndrome, psychiatric*

INTRODUCTION

Mitochondrial disorders (MIDs) are usually multisystem diseases (mitochondrial multi-organ disorder syndromes [MIMODS]) already at onset or become MIMODS during the disease course.^[1] One of the organs frequently involved in MIDs is the cerebrum.^[2] Cerebral involvement may manifest with or without structural abnormalities on cerebral imaging.^[2] Central nervous system

involvement, in the absence of structural cerebral abnormalities, include psychiatric abnormalities (autism, psychosis, confusional state, depression, anxiety, mild cognitive impairment, and dementia) or neurological abnormalities (migraine, migraine-like headache, cluster headache, epilepsy, spasticity, ataxia, Parkinson syndrome, tremor, dysarthria, and dystonia).^[2] Malingering and factitious disorder

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How to cite this article: Finsterer J, Stefan L. Malingering and factitious disorder (Münchausen-syndrome) can be mitochondrial. *Indian J Psychol Med* 2016;38:348-52.

Access this article online

Website:
www.ijpm.info

DOI:
10.4103/0253-7176.185947

Quick Response Code



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(Münchausen-syndrome) has not been reported as manifestation of a MID.^[3]

CASE REPORT

The patient is a 46 years-old, HIV-negative, Caucasian female from nonconsanguineous parents, height 140 cm, weight 52 kg, with a history of migraine since childhood, myalgias and muscle cramps since 1990 with consecutive abuse of analgesics, frequent psychogenic, and rare epileptic seizures since 1990, iron-deficient normocytic and normochrome anemia since at least 1994 which did not respond to substitution, hyperlipidemia since at least 1994, malingering and factitious disorder (Münchausen-syndrome) first diagnosed in 1996, hypothyroidism, respectively, low-FT4 syndrome since at least 1996, ptosis since 5/2007, tremor since 4/2011, recurrent benzodiazepine intoxications, and temporary renal insufficiency. In 2009, a port-a-cath system (port) had been implanted and replaced in 5/2011 to facilitate blood drawing and application of intravenous formulations. She inconstantly reported double vision (since 3/1999), hyperhidrosis, xerostomia, easy fatigability, hemihypesthesia, sensory transverse syndrome, headache, and polydipsia. She was smoking and reported multiple allergies. She also pretended to have undergone cataract surgery but the electronic records did not list such a procedure. The family history was positive for diabetes, dementia, hypothyroidism, coronary heart disease with myocardial infarction, arterial hypertension, chronic obstructive lung disease, gonarthrosis, a history of a Wertheim procedure, and hyperlipidemia in her mother, diabetes (one brother and sister of the mother), and sudden cardiac death in her brother. She had one healthy daughter who was living with her father. For seizures she had received carbamazepine, valproic acid, phenytoin, oxcarbazepine, topiramate, and gabapentine in variable dosages, over variable periods, and with questionable compliance. In 2008 she was on a medication with oxcarbazepine 600 mg/day, pantoprazole 40 mg/day, acetyl-salicylic acid 100 mg/day, desloratadine 10 mg/day, iron-sulfate, and morphinsulfate 60 mg/day. In 1/2011 she was on a medication with tramadol 200 mg/day, gabapentin 600 mg/day, pramipexol 0.54 mg/day, trazodone 150 mg/day, and topiramate 100 mg/day. Two months later she reported a medication with gabapentin 300 mg/day, and topiramate 50 mg/day. Temporarily she received erythropoietin for anemia without effect. Antipsychotic medication was repeatedly proposed, but she refused it. She reported that sumatriptan was ineffective during migraine attacks because of vomiting and that she did not tolerate flunarizine. She irregularly took L-thyroxin but was usually euthyreote or showed low-FT4 syndrome.

At age 38 years, she presented with left-sided hemihypesthesia and sore neck muscles exclusively. Neurologic examination at age 41 years revealed right-sided ptosis, double vision and a sensory transverse syndrome Th12 (right) and Th10 (left). At age 42 years neurologic examination revealed bilateral ptosis with right-sided predominance, reduced tendon-reflexes on the upper limbs, and postural tremor. She pretended to have right-sided hemihypesthesia, which started exactly midline. Blood chemical investigations over 20 years (since 1994) revealed episodic hyperCKemia (maximal value: 1276 U/L), permanent hyperlipidemia, iron deficiency, and transferrin elevation [Figure 1], recurrent hyponatremia, recurrent hypokalemia, occasional hypocalcemia and hypophosphatemia [Figure 2], occasional hypoglycemia, temporary mild renal insufficiency, and frequent hypochrome, and microcytic anemia [Figure 3]. Resting lactate was occasionally slightly elevated. Lactate stress test on a cycle ergometer was normal (lactate: 0.8, 1.6, 1.7, 1.6, and 0.8 mmol/L). Repeated screening for toxins in the serum and urine revealed elevated benzodiazepines on various occasions. Urinary calcium excretion during 24 h was reduced to 1.9 mmol/day (n: 2.5-7.5 mmol/day). Cerebrospinal fluid investigation in 2/1999 revealed slightly elevated protein, normal lactate, and normal cell count. 24 h blood pressure-monitoring in 1/2011 was noninformative. Multiple electroencephalographs (EEGs) had been recorded but only in three was paroxysmal activity ever documented. Several cerebral magnetic resonance imagings (MRIs) or cerebral computer tomographies were normal each time. MRI of the pituitary gland was normal. Carotid ultrasound was normal. Electrocardiogram (ECG) was usually normal but occasionally incomplete right or left bundle-branch-block had been recorded. Holter-ECG in 2/2008 was normal. Transthoracic and transesophageal echocardiography in 12/2011 were normal. Gastrosocopy in 7/1998 detected ulcer ventriculi and in 3/2011 eosinophilic esophagitis. Colonoscopy and videocinematography in 3/2011 and abdominal ultrasound in 8/2012 were noninformative. Muscle biopsy in 12/1999 revealed numerous muscle fibres hypo-reactive for oxidative enzymes, sporadic COX-negative fibers, subsarcolemmal accumulation of mitochondria, increase of glycogen, and abnormal mitochondria with cristae proliferation. Biochemical investigations revealed a combined complex I-IV defect. A second muscle biopsy in 3/2008 showed increased membrane-bound and intracellular glycogen and slightly increased amount of lipid-droplets. Mitochondrial volume was increased in some sections. MID was diagnosed. Search for the mtDNA tRNA (Leu) mutations m.3243A>G, m.3271T>C, and m.3252A>G was negative. Within 20 years she had 175 admissions with a mean duration of 3, 5 days [Table 1].

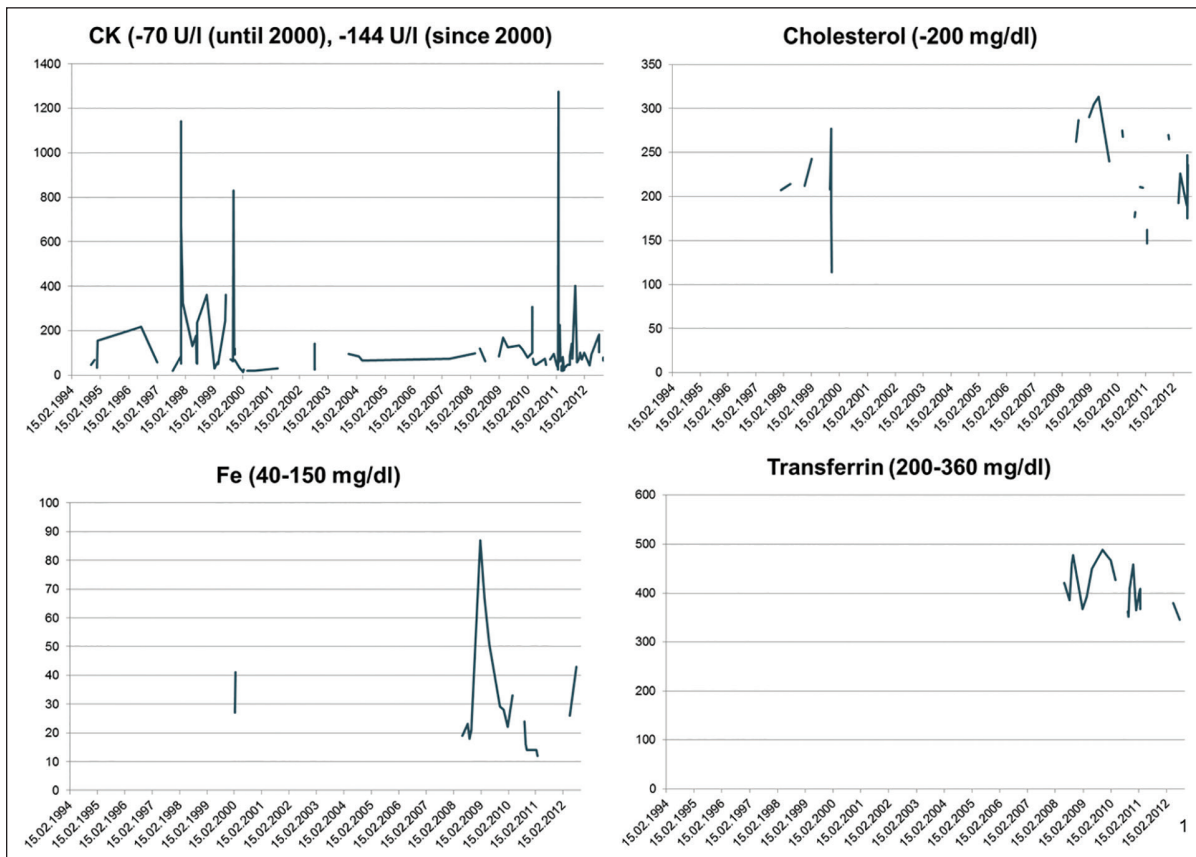


Figure 1: Results of electrolyte determinations (sodium, potassium, calcium, phosphate) in the presented patient over the last 20 years

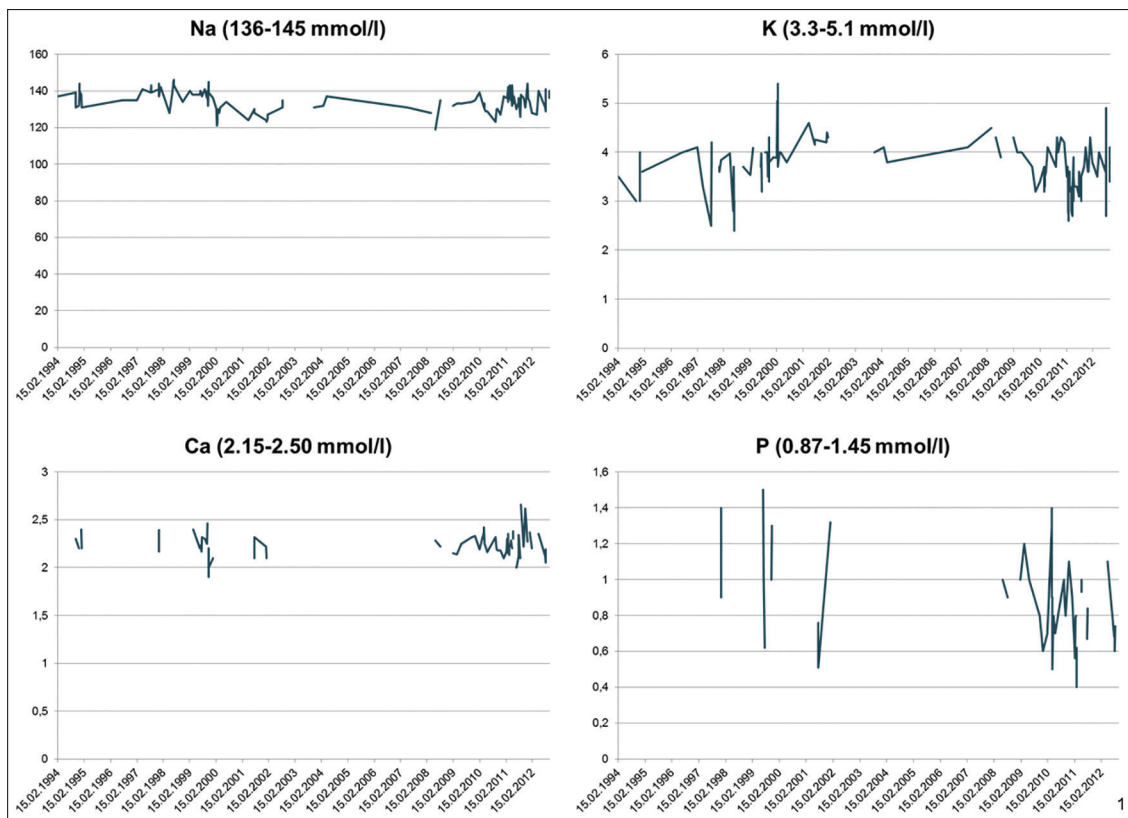


Figure 2: Values of creatine-kinase, cholesterol, serum iron, and transferrin in the presented patient over the last 20 years

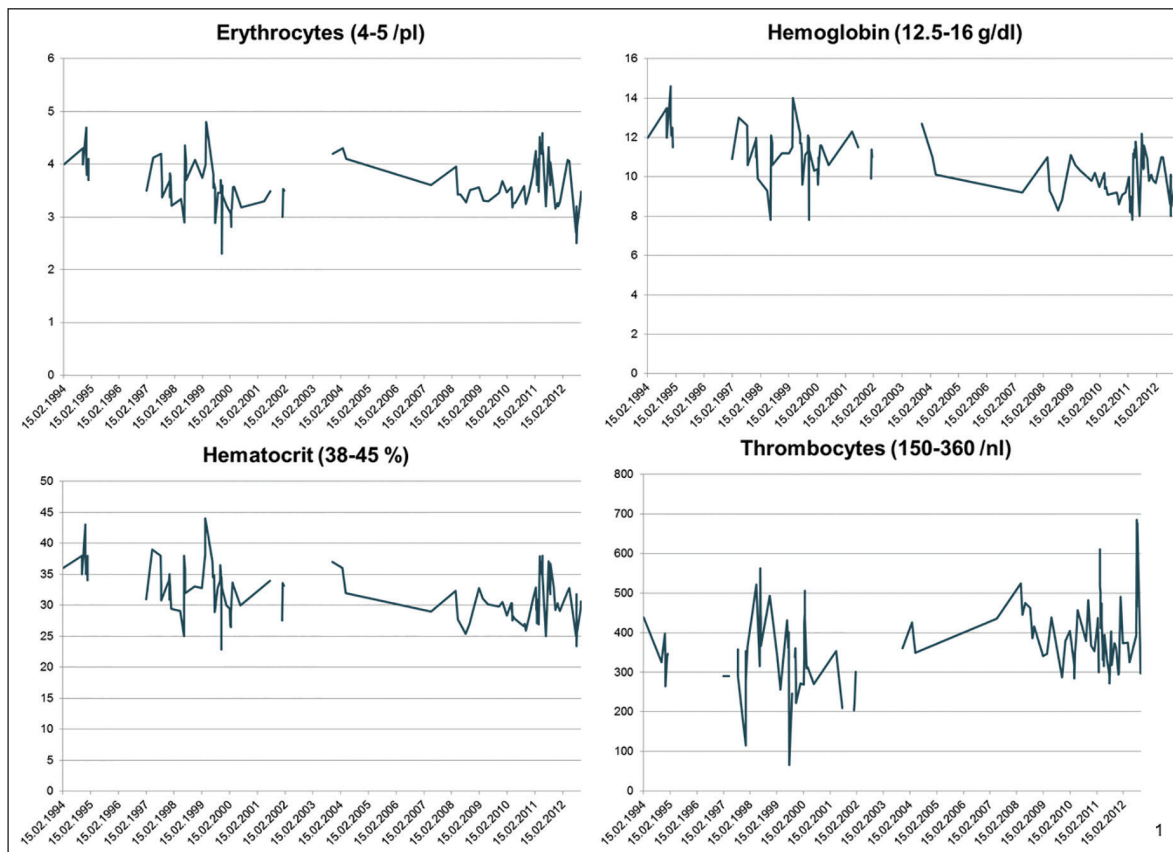


Figure 3: Erythrocyte count, haemoglobin, haematocrit, and thrombocyte counts in the presented patient over the last 20 years

Table 1: Number of admissions, causes of admission, and types of dismissal over a period of 20 years (1994-2014)

Parameter	Total number
Total number of admissions during 20 years	175
Mean duration per admission	3, 5 days
Hospitalizations for 1 day	65
Hospitalizations for 2 days	55
Hospitalizations for 3 days	17
Hospitalizations for 4 days	8
Hospitalizations for 5 days	3
Hospitalizations for >5 days	27
Number of admissions for unknown reasons	108
Admissions due to psychogenic seizures (%)	34/67 (51)
Hospitalizations with psychogenic seizures (%)	19/51 (37)
Number of self-dismissals (%)	27/52 (52)

In 51% the cause of admission was psychogenic seizures [Table 1]. In 37% of the admissions, the patient produced further psychogenic seizures during hospitalisation. In 52%, she was dismissed upon her own request [Table 1].

DISCUSSION

Strong arguments for a MID in the presented patient are short stature, ptosis, reduced tendon-reflexes, tremor, anemia, muscle biopsy, and the family history.^[4] Ptosis is

a key phenotypic feature of MIDs and has been reported in CPEO, MERRF, MELAS, Leigh-syndrome, and nonsyndromic MIDs. Anemia is also a typical feature of MIDs and has been described in Pearson-syndrome, Kearns-Sayre-syndrome, Barth-syndrome, MLASA, XLASA, Leigh-syndrome, LHON, MELAS, TRMA, ARSA, but also in nonsyndromic MIDs.^[5] Short stature is also a frequent phenotypic feature of MIDs and has been reported in syndromic (MELAS, MERRF, MIDD, Leigh-syndrome) and nonsyndromic MIDs.^[6,7] Epilepsy is also a dominant MID manifestation.^[8] Weaker arguments include thyroid dysfunction since hormone levels varied frequently and compliance for intake of L-thyroxine was questionable, and migraine, polydipsia, and arterial hypertension^[9] since her information about headache, intake of drugs, and blood pressure monitoring was assessed as untrustworthy and since arterial hypertension is not generally accepted as a MID manifestation.

Though malingering-disorder has been reported only once as a manifestation of a MID,^[3] it is well-documented that psychiatric disease is frequently part of the phenotypic spectrum of MIDs. Psychiatric abnormalities have been repeatedly reported in association with MIDs and include mood disorder (depression), anorexia nervosa, bipolar disorder,

obsessive-compulsive disorder, cognitive deterioration, psychosis, anxiety, autism, or confusional state.^[3,10] Arguments for malingering-disorder in the presented patient are that she changed her medication frequently, that information about her symptoms and medication was not reliable, that she pretended symptoms, which could not be objectified neither on clinical examination nor on instrumental investigations, that she frequently “produced” psychogenic seizures, resulting in frequent admissions, including intensive care units, and that she accepted painful invasive procedures to get medical attention. An argument for psychogenic seizures was that lactate and prolactin were always normal during or after these events. An argument in favor of epileptic seizures is episodic hyper-CKemia and occasionally paroxysmal EEG activity.

Whether she was injecting insulin via the port to deliberately produce hypoglycemia, or whether she was carrying out blood drawings via the port remains speculative. An argument against voluntary blood drawing is that she had anemia long before the port had been implanted and that anemia was a constant phenomenon over years, making it unlikely that she produced it each time before admission. Arguments against injecting insulin through the port are that she had these attacks already before 2009, that there was never severe hypoglycemia on any of the emergency admissions, that insulin and C-peptide were normal, and that she never presented with typical clinical manifestations of hypoglycemia (temporary tremor, sweating, palpitations, tachycardia, hunger, facial paleness) at any admission. On one of her numerous admissions, she had hypoglycemia and glibenclamide was in her pocket. A further argument for anemia as part of the phenotype is that she had typical MID manifestations, which she could not imitate. Electrolyte disturbances resulted most likely from mild Fanconi syndrome (polyuria, polydipsia, proteinuria, hypophosphatemia, hypoglycemia, and hypouricemia). Myopathy manifested only as ptosis and easy fatigability, cerebral involvement as epilepsy and malingering-disorder, and endocrine involvement as short stature and thyroid dysfunction.

The health system could save costs if patients with psychiatric disease from MIDs are registered and primarily referred to a department equipped to handle MIDs, which is admittedly hardly available. In the light of the increasing prevalence of MIDs, however, institution of departments dedicated particularly to

the management of MIDs would be appreciated, since such units would use their expertise not only for an improved satisfaction of the patient but also for saving public money not only by more specific treatment of these patients but also by relief of the many specialized departments, including neurological departments, to which these patients are referred nowadays but which are frequently overburdened with the diagnosis MID and its plethora of phenotypic expressions.

This case shows that MIDs also manifest with a malingering-disorder but normal cerebral imaging. Management of such patients could be difficult for departments not familiar with MIDs and facilitated and improved if these patients are taken seriously and if they are treated at departments particularly dedicated to the handling of MIDs.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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