Neurological manifestation of systemic diseases

EP4256

Anti-aquaporin 4 autoantibodies in patients with systemic lupus erythematosus without CNS involvement: a long-term assessment

H. Alexopoulos, I. Tatouli, S. Akrivou, A. Tzioufas, M.C. Dalakas

Neuroimmunology Unit, Department of Pathophysiology, National and Kapodistrian University of Athens, Athens, Greece

Introduction: Anti-AQP4 autoantibodies are specific for the Neuromyelitis Optica-Spectrum Disorders (NMOSD). They have been previously described in patients with Systemic Lupus Erythematosus (SLE) who concurrently have neurological signs consistent within NMOSD. Whether these autoantibodies are also present in the sera of non-CNS-SLE patients, is unknown.

Methods: Sera from 89 non-CNS-SLE patients identified by clinical record review were screened for anti-AQP4 autoantibodies by an in-house cell-based-assay (CBA) using M23-AQP4-transfected cells. Seropositivity was confirmed by a commercial CBA assay. Archived samples from seropositive patients, obtained over a 10-year period were also analyzed.

Results: Two out of 89 (2.2%) of non-CNS-SLE patients were anti-AQP4 seropositive. Archived samples (7 and 16 from each patient, dating back 8 and 10 years, respectively) were also AQP4-positive as confirmed by both assays. These AQP4-positive SLE patients are women with disease duration of 16-17 years, a mild type of renal involvement and a wide spectrum of other autoantibodies. Both patients had never exhibited any CNS-related symptoms based on neurologic examination. A current brain and spinal cord MRI did not reveal any NMOSD-compatible lesions.

Conclusions: AQP4-antibodies are present in some SLE patients and persist for many years, even without developing clinical or MRI signs of NMOSD. It is a possible that the antibodies in these patients result from the underlying polyclonal B cell activation of the immune system, typical for SLE, and may not be pathogenic. Alternatively, the possibility that the antibodies might herald the potential of developing CNS disease even after a decade, cannot be excluded.

Disclosure: Nothing to disclose

EP4257

Abstract withdrawn

EP4258

Cerebral vasculitis: a report of 17 cases

Y. Cherif¹, S. Younes¹, H. Haj Kacem¹, O. Berriche¹, H. Hamza², S. Jerbi², M.H. Sfar¹

¹Internal Medicine and Endocrinology, ²Radiology, Tahar Sfar University Hospital, Mahdia, Tunisia

Introduction: Cerebral vasculitis (CV) is rare. They may be primary or related to systemic, infectious or neoplastic diseases.

Methods: Retrospective study of 17 cases of CV. All patients underwent a neurological and ophthalmological examination, cerebral imaging and immunological analysis.

Results: There were 11 men and 6 women, whose mean age: 56 years (25-75 years). The history was noteworthy with arterial hypertension in 1 patient, diabetes in 2 patients, pulmonary embolism in 1 patient, retinal vasculitis in 1 patient and meningioma in 1 patient. The CV was diagnosed at primarily motor impairment in 6 patients, headache in 4 patients, dementia in 2 patients, intracranial hypertension in 2 patients, coma and in 1 patient and psychiatric disorders in 2 patients. Five patients had generalized tonic-clonic seizures. All patients had T2 hyperintensities in the periventricular white matter in 13 cases, in the brainstem in 2 cases and in the cerebellum in 2 cases. CV was secondary to Behçet’s disease in 5 cases, Sjogren’s syndrome in 4 cases, giant cell arteritis in 3 cases, systemic lupus erythematosus in 2 cases, antiphospholipid syndrome in 2 cases and Susac syndrome in 1 case. All patients were treated with high dose corticosteroids regimen, associated with Cyclophosphamide in 2 cases, Azathioprine in 2 cases, anticoagulant drugs in 3 cases and antiplatelet drugs in 13 cases. The outcomes were quite favorable with complete or partial recovery of neurological deficit in 16 cases and disappearance of psychiatric disorders in 1 patient.

Conclusions: We present different clinical features of CV, main causes, its management and prognosis. The diagnosis is held on a basis of clinical, biological and MR findings even if the definite diagnosis is based on cerebral biopsy.

Disclosure: Nothing to disclose
EP4259

A case series of giant cell arteritis: vascular risk factors, manifestations and diagnostic evaluation

A. Costa¹², A. Silva-Pinto¹, P. Abreu¹²
¹Neurology, Centro Hospitalar São João, ²Faculty of Medicine, University of Porto, ³Infectious Diseases, Centro Hospitalar São João, Porto, Portugal

Introduction: The giant cell arteritis (GCA) is the most common vasculitis after the age of fifty. It is more frequent in the female gender. The mainstays of GCA diagnosis are: clinical judgment, exclusion of other diagnosis and temporal artery biopsy.

Methods: We conducted a retrospective, transversal study of patients with GCA hospitalized between 2006 and 2012. The aim of this study was to characterize the hospitalized population with GCA concerning vascular risk factors, manifestations of the disease, and diagnostic evaluation.

Results: We included 30 patients with ACG. The mean age at the diagnosis was 74.8 years and the female:male ratio was 2:1. Hypertension was present in 70%, hyperlipidemia in 40%, diabetes in 20% of patients and 17% were smokers. Five (17%) patients had a stroke as first manifestation of GCA. The classification criteria (American College of Rheumatology 1990) were fulfilled in 87% of patients. Fourteen temporal artery biopsies were performed and 79% had compatible alterations. Forty-five percent of the twenty-two temporal artery Doppler ultrasound done had compatible alterations with GCA.

Conclusions: The mean age and the gender ratio are similar to other series of GCA. The higher prevalence of stroke comparing to other series may be due to the high prevalence of vascular risk factors or due to a selection bias (hospitalized patients with possible more severe disease). Temporal artery biopsy is still considered the gold standard exam to diagnose GCA, nevertheless Doppler ultrasound is an high specific, easy, accessible and non invasive exam that may help in its diagnose.

Disclosure: Nothing to disclose

EP4260

Functional disorders in the neurology ward

C. Fernandes, N. Ferreira
Neurology Department, Hospital Garcia de Orta, Almada, Portugal

Introduction: Functional disorders are frequent in neurological practice, placing diagnostic and therapeutic uncertainty. We aim to characterize a group of patients with neurological functional disorders and determine prognostic factors.

Methods: Retrospective study in patients admitted to neurology ward with final diagnosis of functional disorder, in five years. Demographics, clinical, investigation, therapeutics and evolution were analysed.

Results: Seventy patients identified: 71.4% female, mean age 43.2 years, admitted mainly from the Emergency Room, 35.7% had history of psychiatric disorder and 17.1% neurological. Most had acute presentation (51.4%≤24h; 77.1%≤1week) and 65.8% with multiple symptoms, predominantly sensory deficit and paresis (42.9%). Neuroimaging (98.6%) and neurophysiological (41.4%) examinations prevailed in the investigation. Only 22.9% had psychiatric evaluation. At discharge, 37.5% maintained symptoms and 27.1% had complete resolution. Psychotropics were prescribed in 80%. Inconsistencies in neurological examination (27.1%) and findings incongruous with organic lesion (12.9%) chiefly supported the diagnosis. At discharge, 41.4% were referred to neurology outpatient, 27.1% to psychiatry, 15.7% to both. With mean follow-up of 866.4 days in 54 patients, 27.8% maintained symptoms or had recurrences, only two were readmitted. In none the diagnosis changed to organic disorder. No association was found between prognosis and gender, previous psychiatric or neurological disorder, evolution time, clinical status at discharge, psychotropic therapy or specific outpatient referral.

Conclusions: The approach to these patients was diverse. Despite the favourable outcome in most, symptoms persisted or recurred in many and prognostic factors were not identified. The establishment of multidisciplinary follow-up and effective therapeutic strategies is warranted.

Disclosure: Nothing to disclose
**EP4261**

**Multifocal motor neuropathy associated with infliximab treatment**


**Introduction:** Multifocal motor neuropathy (MMN) is an immune-mediated disorder characterized by motor-conduction block in nerve-conduction studies. Infliximab is a TNF blocker which is used to treat inflammatory diseases. Several immune-mediated conditions have been reported as adverse events of infliximab use.

**Methods:** We report 2 cases of MMN associated with the use of infliximab.

**Results:**

Case A: A 24-year-old male presented with a slowly progressive asymmetrical weakness of the arms, without sensory loss. He has been on treatment with infliximab for Crohn’s disease for 3 years. Clinical examination revealed signs of lower-motor-neuron disease. A complete diagnostic workup was performed. Conduction studies showed motor conduction blocks in several nerves of both upper and lower limbs.

Case B: An 82-year-old female, with Crohn’s disease, has a chronic mobility problem that was strongly exacerbated when she was started on treatment with infliximab 4 months before consultation. Clinical examination showed diffuse pyramidal signs in relationship with her prior condition, but also revealed flaccid and areflexic paraplegia. A complete diagnostic workup was done. Conduction studies revealed motor conduction blocks in several nerves of her legs.

In both cases, infliximab was withdrawn and treatment with intravenous immunoglobulin was established.

**Conclusions:** The use of TNF-alfa blockers has been linked to a variety of neurological immune-mediated disorders as adverse effect. Withdrawing the offending agent and immunomodulatory therapy may be required to improve clinical outcome. MMN is a condition that has rarely been linked before to infliximab.

**Disclosure:** Nothing to disclose

**EP4262**

**Neurologic complications after bariatric surgery**

J.K. John, J.Y. Al-Hashel, A. Rady, V. Periasamy

**Introduction:** Obesity is a major public health problem worldwide and the concern about obesity and diseases related to that is also increasing. Hence an increasing number of patients undergo various bariatric surgical procedures. However these procedures can lead to many complications and those affecting the nervous system are often particularly disabling. We describe the neurologic complications observed among 21 patients who underwent bariatric surgery.

**Methods:** We describe the neurologic complications following bariatric surgery in 21 patients who were seen in a tertiary referral Neurology Centre in Kuwait during a period of 3 years from 2010-2013.

**Results:** The most common neurological complication observed was painful peripheral neuropathy. Other manifestations include optic neuritis, central demyelination, polyradiculoneuropathy, myelopathy and myopathy. One patient who had been on treatment for multiple sclerosis (MS) developed rapid deterioration of disability and progression of the disease following bariatric surgery. Another patient had acute polyradiculoneuropathy and subsequently developed MS. Vitamin D deficiency was the most common nutritional deficiency detected and some had B12 and one patient had copper deficiency. There was no definite correlation of any specific nutritional deficiency with the neurologic complications.

**Conclusions:** A wide spectrum of neurologic complications can occur following bariatric surgery. Proper pre-operative and post operative evaluation with intensive nutritional management and frequent follow up assessment is necessary to reduce these complications.

**Disclosure:** Nothing to disclose
EP4263

**Encephalopathy and neuroimaging in acute porphyria**

E. Pischik\(^1\), R. Kauppinen\(^2\)

\(^1\)Department of Neurology, Consultative and Diagnostic Center with Polyclinics, Saint Petersburg, Russian Federation, \(^2\)Department of Medicine, Research Program in Molecular Medicine, Biomedicum-Helsinki, University of Helsinki, Helsinki, Finland

**Introduction:** The aim of the study was to characterise a spectrum of clinical manifestations and neuroimages in patients with encephalopathy and acute porphyria.

**Methods:** Brain MRI/CT was performed in ten well-characterised patients with acute intermittent porphyria (AIP, confirmed biochemically and genetically) in various stages of encephalopathy.

**Results:** Acute encephalopathy manifested as severe mental symptoms and signs of focal CNS involvement such as seizures, Babinski signs, hemiparesis or ataxia. In all patients, acute encephalopathy was a sign of an acute attack. Other symptoms included abdominal pain (n=8), dysautonomia (n=10), acute motor neuropathy (n=7) and at least 10-fold elevation of urine porphobilinogen. In two patients, focal lesions corresponded posterior reversible encephalopathy syndrome (PRES) when brain MRI/CT were performed soon after seizures. In a patient with syndrome of inappropriate antidiuretic hormone secretion (SIADH), the bright signal from the neurohypophysis was reduced. In other seven patients, brain MRI/CT was normal.

**Conclusions:** Seizures can predict PRES if neuroimaging was taken within a day. Focal symptoms were transient and no residual MRI lesions could be detected. The frequency of reversible brain oedema in AIP is probably underestimated since it may be short-lasting and often indistinguishable on MRI. Mental symptoms even though severe, caused no abnormalities in brain MRI. PRES could partly explain the pathogenesis of encephalopathy in acute porphyrias. However, the primarily mechanisms disrupting blood-brain barrier, thereafter permitting access of neurotoxins (porphyrin precursors) into the CNS, are unknown. SIADH during an acute attack may result from an inappropriate release rather than an inappropriate synthesis of ADH.

**Disclosure:** Nothing to disclose

EP4264

**Treatment of an acute attack with severe neurological manifestations in patients with acute intermittent porphyria (AIP): haem arginate vs. glucose**

E. Pischik\(^1\), R. Kauppinen\(^2\)

\(^1\)Department of Neurology, Consultative and Diagnostic Center with Polyclinics, Saint Petersburg, Russian Federation, \(^2\)Department of Medicine, Research Program in Molecular Medicine, Biomedicum-Helsinki, University of Helsinki, Helsinki, Finland

**Introduction:** Currently, no evidence based data of benefit for haem treatment vs. glucose to achieve neurological recovery in patients with acute intermittent porphyria (AIP) manifesting neurological deficits exists.

**Methods:** 14 AIP patients with a severe acute attack, complicated by peripheral neuropathy (PNP, n=12) and/or encephalopathy (n=3).

Acute attacks were treated with 20% glucose-infusions (300-500g/day, 5-12 days, n=7) or haem arginate (Normosang\(^®\), 2.5-3mg/kg, 4 days, Orphan Europe, n=7) after the diagnosis of AIP was confirmed (6-52 days after the onset of an attack). The choice of treatment was based on availability of haem arginate solely.

**Results:** The plateau phase was detected significantly earlier in patients treated with haem than those treated with glucose (7.0±2.4 vs. 10.8±3.1 days, p=0.035). Minimal improvement of muscle weakness could be recorded a week after the last infusion. In two cases, progression of neurological deficit required additional infusions of haem resulting in a second plateau phase in one case, but not in another, who died due to septicemia. Haem arginate infused 1-4 days after the onset of recurrent attacks has prevented neurological deficits (n=8). In contrast, the patient treated with 20% glucose 4 days after the onset of new acute attack, experienced recurrent PNP.

**Conclusion:** Haem arginate is more efficient than glucose in achieving the plateau phase for progressing neurological deficits in AIP. This allows to shorten the duration of mechanical ventilation, activate patients and to prevent complications of an attack. There is no clear benefit of haem in stable or self-resolving neurological deficits.

**Disclosure:** Nothing to disclose
EP4265

Acute transverse myelitis following stem cell transplantation for haematological malignancies: a series of three cases

K. Su1, R. Brown1, R. Dooley1, S. Alimam2, A. Zermansky1, D. McKee1

1Department of Neurology, Greater Manchester Neurosciences Centre, 2Department of Haematology, Manchester Royal Infirmary, Manchester, United Kingdom

Matched unrelated haematopoetic stem cell transplantation (HSCT) is a high risk but potentially curative treatment increasingly used for a range of haematological malignancies. Following the procedure, a number of neurological problems can arise, including opportunistic infections, complications of bone marrow suppression, relapse of the haematological disease with neurological involvement, and a number of dysimmune phenomena such as polymyositis and inflammatory polyneuropathies. We report a series of three cases of acute transverse myelitis (ATM) following HSCT. All were male with age ranges between 46 and 55. Two had a diagnosis of Acute Myeloid Leukaemia and one Acute Lymphoblastic Leukaemia. The first neurological symptom developed between 86 and 195 days post HSCT. In each case the presentation was of a rapidly progressive myelopathy clinically typical of ATM. All had high signal lesions within the spinal cord on MR imaging, and in two of the cases the abnormality met the radiological criteria for longitudinal extensive transverse myelitis. In each case the patients were extensively investigated to exclude an infectious or malignant aetiology for ATM. None of the patients were found to have Aquaporin 4 antibodies in the serum. A combination of treatments with steroids, plasma exchange and intravenous immunoglobulin were given, and there have been variable degrees of improvement up to full recovery. Although there are other cases in the literature, this is the largest series of post HSCT myelopathy yet reported. Neurologists who deal with haematology units need to be aware of this syndrome and its important differential diagnosis.

Disclosure: Nothing to disclose

EP4266

Neurological disorders in children with ichthyosis: lessons from eight patients diagnosed with Sjögren-Larsson syndrome and a patient with lamellar ichthyosis in a rural hospital

A. Vural1, S. Saral2

1Department of Neurology, 2Department of Dermatology, Besni Government Hospital, Adiyaman, Turkey

Introduction: Ichthyosis is a dermatological disorder with more than 28 subtypes. Some rare congenital forms like Sjögren-Larsson Syndrome (SLS) may be accompanied by neurological symptoms. SLS is a very rare (1/200,000), autosomal recessive disease and is diagnosed clinically when the classic triad including congenital ichthyosis, mental retardation and spastic paraparesia is seen in a child. In addition, other forms of ichthyosis with severe dermatological problems may rarely cause vitamin D deficiency and hypocalemia presenting with neurological manifestations.

Method: We present clinical, laboratory and neuroradiological findings of eight patients with SLS and one patient with severe lamellar ichthyosis who are all diagnosed in a rural hospital in Turkey and discuss neurological manifestations seen in patients with ichthyosis.

Results: Four male and four female patients (aged between 2 and 19 years) with ichthyosis, mild to moderate mental retardation and severe spastic paraparesis were diagnosed clinically with SLS. All patients except one were from the same kindred. All patients had mild skeletal deformities, two patients had epilepsy and one patient had whole body tremor, additionally. Brain magnetic resonance imaging of five patients revealed symmetrical, periventricular white matter lesions which is characteristic for SLS. Ninth patient with severe lamellar ichthyosis applied to neurology clinic with moderate to severe muscle weakness and cramps. Hypocalcaemia, hyperphosphatemia and high alkaline phosphatase levels were detected. His symptoms recovered after replacement therapy.

Conclusion: Although skin appearance attracts most of the attention, it should be kept in mind that several neurological manifestations may also be seen in children with ichthyosis.

Disclosure: Nothing to disclose
EP4267

Neuro-Behçet syndrome with spinal cord involvement

B. Zeydan¹, U. Uygunoglu¹, S. Ugurlu², E. Seyahi², S. Saip¹, A. Siva¹
¹Department of Neurology, 2Department of Internal Medicine, Division of Rheumatology, Istanbul University Cerrahpasa School of Medicine, Istanbul, Turkey

Introduction: Central nervous system parenchymal involvement is named as parenchymal neuro-Behçet syndrome (NBS) or intra-axial NBS. Neurological manifestations are usually related to brainstem. In comparison to brainstem lesions, spinal cord involvement (SCI) is rarely seen in parenchymal NBS. It is reported that the prevalence of spinal NBS ranges between 2.5 to 30%. We examined 19 NBS patients with SCI to determine clinical aspects and course of disease in this subgroup.

Methods: Nineteen patients were included in the study who attended our NBS center and were diagnosed as NBS myelitis. Clinical features and outcomes of patients were evaluated.

Results: 14 patients were men, five were women. Behçet Syndrome and NBS mean age at time of diagnosis were 24.2±6.1 and 25.5±6.6 respectively. Most patients were presented with paraparesis and/or sphincter dysfunction evolving over time and four had more than one spinal attack. Accordingly spinal cord MRIs revealed single/multiple, mostly long segment, cervical and/or dorsal lesions or spinal cord atrophy. Some patients had normal cranial imaging, others had additional cranial involvement, mainly brainstem lesions. Intravenous methylprednisolone followed by oral corticosteroids were administered for myelitis attacks. Azathioprine was the first choice of long-term treatment, in more severe or recurrent myelitis, infliximab was another option. After ischemia, apart from immunosuppressive therapy (azathioprine and/or corticosteroid and/or colchicine), nine patients had acetylsalicylic acid (ASA), two patients had ASA-dipyridamol and one had warfarin treatment. Average follow-up was 77 months, average EDSS was 5.2.

Conclusions: Lesions in NBS myelitis are usually relatively long, located in the center of the cord with peripheral edema. Differential diagnosis is important, since NBS myelitis associated with disability is recognised in general as a poor prognostic factor.

Disclosure: Nothing to disclose

EP4268

A subgroup of neuro-Behçet syndrome: intracranial arterial involvement

B. Zeydan¹, U. Uygunoglu¹, M. Tutuncu¹, C. Yalcınkaya¹, A. Altintas¹, S. Ugurlu², E. Seyahi², S. Saip¹, A. Siva¹
¹Department of Neurology, 2Department of Internal Medicine, Division of Rheumatology, Istanbul University Cerrahpasa School of Medicine, Istanbul, Turkey

Introduction: Parenchymal neuro-Behçet syndrome (NBS), the most common type of NBS is seen as a result of small-vessel vasculitis mainly with brainstem lesions. In non-parenchymal NBS, involvement of large vessels and dural sinus thrombosis are seen. We report 15 NBS patients with cranial arterial involvement (CAI) to determine clinical-radiological features of CAI in NBS which is extremely rare.

Methods: Fifteen patients who attended our NBS-center between 1994-2013 were diagnosed as NBS with CAI. Clinical features, risk factors (RF), prognosis were examined.

Results: All patients were men. Behçet Syndrome and NBS mean age at time of diagnosis were 31.5±9.5 and 37.6±13.1 respectively. One patient had hypertension, one had diabetes, one had hyperlipidemia as stroke RF. One patient had migraine, one had pulmonary-artery aneurysm, six patients were smoking, one had history of opioid use. The most frequent onset symptom was hemiparesis. 13 patients had middle cerebral artery infarction (AI), two had posterior cerebral AI, one had superior cerebellar AI. Echocardiography-cranial MR angiography examinations were normal except one patient had cardiac-septal hypokinesia and one had superior cerebellar artery aneurysm. After ischemia, apart from immunosuppressive therapy (azathioprine and/or corticosteroid and/or colchicine), nine patients had acetylsalicylic acid (ASA), two patients had ASA-dipyridamol and one had warfarin treatment. Average follow-up was 67 months, modified-Rankin-Scale scores before and after treatment were 1.7 and 0.8 respectively.

Conclusions: Venous inflammation is the main neurological involvement in BS patients, whereas intracranial arterial involvement in NBS is rare, but it can occur during the course of disease independent of other stroke RF.

Disclosure: Nothing to disclose
EP4269
Nocturnal seizures: a misdiagnosis of hypoglycemic episodes
A. Lopez, A. Hernández, C. Valencia, N. Giraldo, J.J. Bravo, S. Carrasco
Universitary General Hospital, Ciudad Real, Spain

Introduction: Hypoglycemia is commonly diagnosed in emergency departments. Nevertheless, this diagnosis may be a challenge when sympathoadrenal manifestations go unnoticed and neuroglycopenic symptoms prevail; moreover, glycemia measurement may be normal at the time of the evaluation. We report two cases who presented neuropsychiatric symptoms suggestive of nocturnal seizures, related to severe hypoglycemia.

Methods: A 40-years-old type I diabetic patient, treated with insulin for five years, reported recurrent episodes of confusion, automatisms, optical illusions and uncontrolled laugh, occurring always during the early morning hours. They lasted from 10 to 15 minutes being followed by drowsiness. A 50 years old man exhibited bizarre behaviour at morning, with uninhibited and infantile conducts, always detecting normal glycemia levels at the emergency room. MRI and repeated EEG were normal in both cases.

Results: During admission, morning glycemia of 39 mg/dl was detected in the first case, and after insulin adjustment the symptomatology was solved. In the second case, a tomography performed several weeks later, showed a pancreatic insulinoma. After distal pancreatectomy symptoms did never recur.

Conclusions: The most frequent causes of hypoglycemia are sulphonilureas or insulin; other include alcohol, Addison’s disease or insulinoma, which is the most common cause of endogenous hyperinsulinemic hypoglycemia. In large series, 20 percent of patients with unknown insulinoma had been misdiagnosed of a neurologic or psychiatric disorder. Serial determinations of glycemia are mandatory in the evaluation of a patient with any behavioural disorder or confused state, as hypoglycemia can mimic many neurologic disorders, including stroke or epilepsy.

Disclosure: Nothing to disclose

EP4270
Neuro-Wilson’s disease – about seven cases and review of the literature
L. Ben Algia, I. Chatti, M. Benhalima, Z. Saied, A. Khefifi, S. Ben Amor, S. Benammou
Department of Neurology, CHU Sahloul, Sousse, Tunisia

Introduction: Wilson’s disease is an autosomal recessive disorder of copper overlap, dominated by neuropsychiatric and hepatic symptoms. The aims is to to review the genetic aspects, diagnosis and treatment of Neuro-Wilson through a series of patient followed in the neurology department sahloul, CHU Sousse.

Methods: We report seven cases collected in the department of neurology CHU Sahloul Sousse. All patients had neurological signs with or without extraneurological symptoms. The diagnosis of Wilson’s disease was based on clinical, biological and radiological result.

Results: Seven patient, two boys and five girls. The mean of age was 25.5 with an extreme ranging from 14 years to 48 years. The neurological signs were revealing the disease in six patients and the most common symptoms were tremor. One patient had cerebellar ataxia and another has had seizure. Three of patients had a familial form with an autosomal recessive transmission. The ring of Kayser Flecher was found in one patient. Cupric balance was disturbed in all patients including one associated hemochromatosis. The cerebral MRI was pathologic in two patients with lesions in basal ganglia. All patients were treated by D penicillamine with good evolution in five patients.

Conclusions: When left untreated, the evolution of Wilson’s disease is always fatal. Treatment is based on the chelating copper, zinc salts and liver transplantation . The prognosis of Wilson’s disease appears even better than the neurological and liver symptoms are not pronounced.

Disclosure: Nothing to disclose

EP4271
Abstract withdrawn
Neurological manifestations of acute intermittent porphyria (AIP)

E. Pischik¹,², R. Kauppinen²
¹Department of Neurology, Consultative and Diagnostic Center with Polyclinics, Saint Petersburg, Russian Federation, ²Department of Medicine, Research Program in Molecular Medicine, Biomedicum-Helsinki, University of Helsinki, Helsinki, Finland

Introduction: Acute peripheral neuropathy (PNP) and/or encephalopathy may occasionally develop in attack of acute intermittent porphyria (AIP). Most of the studies on neurology in porphyria were published 40-60 years ago, before the DNA-diagnostics of AIP allowing the precise diagnosis and neuroimaging became available.

Methods: 18 AIP patients with severe peripheral neuropathy and/or encephalopathy studied clinically, by neuroimaging and electroneuromyography prospectively during an acute attack and in remission (1996-2013).

Results and conclusions: PNP or encephalopathy are signs of a complicated acute attack and usually iatrogenic since they mainly develop due to administration of porphyrinogenic drugs used when the diagnosis of acute porphyria was not considered.

The major pattern of PNP associated with abdominal pain, dysautonomia, CNS involvement and mild hepatopathy could be demonstrated. If more strict biochemical criteria (>10-fold increase in excretion of urinary porphobilinogen) are applied, neurological manifestations of porphyria are probably more homogeneous than described previously, which suggests that some of the neurological patients described previously may have secondary porphyrinuria rather than acute porphyria.

Based on nerve conduction studies, PNP is primarily axonal. However, in severe cases definite features of demyelination appear early in the course of PNP.

Porphyric encephalopathy can be visualized as a posterior reversible encephalopathy syndrome (PRES) if MRI performed within a few hours from the onset manifested with seizures.

Currently the prognosis of neuropathy and encephalopathy in acute porphyria is good even in severe attacks, but physicians should be aware of a potentially fatal outcome of the disease.

Disclosure: Nothing to disclose