Association of British Neurologists

Self-Assessment Questions for consultants & trainees 2015

After a brief hiatus, it's back, and we enclose 10 more questions to educate, delight, infuriate and antagonise you all. There are no official CPD points, but you can self certify, and we encourage you to do so. I suggest that if done properly, this should account for about 5 to 6 hours work, including reading around the subjects.

Ideally, we encourage you to complete all 10 questions via SurveyMonkey link before downloading the answers pdf. Once you have completed the questions, you can access a CPD certificate.

We are very grateful to all of you who submitted questions, and if we did not use your question this time round, it may well appear in future exercises. And for those of who feel the questions are too dull/pedantic/hard/easy/wrong/bizarre, then put your typing where your mouth is and send me a question or two, we are always in need! All comments welcome.

Richard J Davenport May 2015

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TEC Chair (on behalf of TEC)

Contents

Question 1

You assess a 65-year-old lady in the intensive care unit. She was admitted to the local hospital having been found confused at her home by her daughter. On admission, she was found to be confused with a GCS of 14 (E4 V4 M6). Pulse rate 84/min, BP 136/84 mmHg, temperature 37.5^{o}C. She was moving all four limbs, with no reflex asymmetry and down-going plantar responses. In the ED, her consciousness deteriorated, with the GCS dropping to 5 (E2 V2 M1). She was intubated and transferred to the intensive care unit.

The past medical history was unremarkable, except for depression for several years, treated with Fluoxetine. A few days prior to admission, she was noted to be hallucinating and was assessed by the general practitioner. A psychiatric assessment was done and a diagnosis of psychosis was made. She was offered an inpatient psychiatric treatment, but the family refused, opting to take Mirtazepine as an outpatient. She made some improvement with this, although there had been episodes of slow involuntary movement of the hands and face, attributed to medication.

On examination in ITU she is intubated and ventilated, but sedation has been weaned. She has no spontaneous movements, but seems to be responding to commands with flickering of eye movements. There is some rigidity, with preserved reflexes and down going plantar responses. The ITU nurse tells you that there has been some fluctuation of blood pressure and heart rate, not requiring any pharmacological intervention.

Investigations:

Hb: 12.6 g/dL, white cell count: 7.6x10^{9}/L, platelets: 350x10^{9}/L
Na: 135 mmol/L, K: 4.1 mmol/L, Urea: 6.1 mmol/L, Creatinine: 45 µmol/L, Albumin: 30 g/L, Bilirubin: 5 µmol/L, AST: 16 U/L and CRP: 12 mg/L.
CT head scan: unremarkable
EEG: paroxysmal slow waves in both hemispheres with no clear epileptiform activities.
CSF: normal opening pressure, white cells 5 (all lymphocytes), protein 0.48 g/L, glucose 4.2 mmol/L (blood glucose 6 mmol/L).

Which of the following tests is most likely to inform the diagnosis?

Question 1 Answers

A. ASO (streptococcal) serology/titres
B. Caspr2 antibodies
C. MR head
D NR1 subunit antibodies
E. Pelvic ultrasound

Suggested Answer

D

Explanation / Comments

This is an acute encephalopathic presentation, preceded by a psychiatric history. There are no clear clinical or laboratory evidence of acute infection. In practice, most patients with this presentation might be given empirical treatment, at least with aciclovir until initial results are available. This patient has a history suggestive of psychosis with some involuntary movements. Examination shows rigidity, with possible catatonia. Although facio-brachial dystonic seizures are described with LGI-1 (one of the targets for VGKC-complex) antibodies, the description of the involuntary movements is more likely to represent choreo-athetoid movements described in NMDAR encephalitis; antibodies are mainly directed against the NR1 subunit. Although post-streptococcal chorea should be considered, this is unlikely with the history of psychiatric symptoms and respiratory failure. Hyponatremia is seen in LGI-1 (VGKC-complex) autoimmune encephalitis, but is not a manifestation of NMDAR encephalitis.

NMDAR encephalitis is now thought to have a two stage process. The initial stage includes psychosis, seizures, confusion and memory loss. Ten to 20 days later, most patients develop basal ganglia and brain stem changes in the form of involuntary movements, reduced consciousness and autonomic disturbances. CSF lymphocytic pleocytosis may be seen early in the disease course, with 30% of patients showing raised CSF protein and oligoclonal bands later in the disease. Early reports suggested an exclusive association with ovarian teratoma, but as the phenotype has expanded, paraneoplastic aetiology seems less common (less than 30-50%). However, a thorough tumour screen is required in all patients with confirmed NMDAR antibodies. Immunotherapy early in the disease course is associated with a favourable prognosis, however most patients require more than three courses and the disease is known to relapse frequently (at least 25% in non-tumour patients), unlike the VGKC-complex LE which is often a monophasic illness.

Caspr2 antibodies are often seen in neuromyotonia, although rarely can be seen in patients with limbic encephalitis. $GABA_A$ antibodies have been recently described in serum and CSF of patients with encephalitis with refractory seizures and status epilepticus.

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Acknowledgements / Consent (IF APPLICABLE)

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Question 2

A 67 year old man was referred after noting mild weakness in his right hand 9 months previously; he had seen an orthopaedic surgeon who considered carpal tunnel syndrome but a steroid injection for this had made no difference. Subsequently the patient noted difficulty in taking his wallet from his back pocket.

He volunteered no other symptoms. On direct questioning he had tripped once in the previous month. He had seen his GP with some difficulty in swallowing 20 months previously, but this was not investigated, and had caused fewer problems recently as he avoided dry food such as toast.

He had travelled to see his son working in Ouagadougou 3 months before and had suffered a coryzal illness on his return. His paternal uncle and great-uncle had died of Duchenne muscular dystrophy in their teenage years. He was taking Stemetil (since an episode of vertigo 2 years previously), nifedipine (Raynaud's phenomenon) and citalopram (low mood). Ten years previously he had consulted a rheumatologist with possible rheumatoid arthritis and had taken prednisolone for a few months.

Examination showed his walking to be slow but no other parkinsonian signs. Cranial nerve examination showed no abnormality except for some drooping of his lower lids and difficulty puffing his cheeks out. On drinking a glass of water he coughed.

Muscle bulk, tone, coordination and reflexes were normal except for some thinning of the right forearm. No contractures or myotonia were evident. Sensory examination was normal. Neck and proximal arm strength was normal, but it was possible to overcome his grip in both hands, more easily on the right, and mild weakness of right wrist flexion. He could not rise from a kneeling position without using his hands, but strength in his legs was normal when tested on the couch.

GP routine bloods were normal apart from a CK of 320 (<200).

What is the most likely diagnosis?

Q2 Answers

A Dystrophinopathy

B Inclusion body myositis
C Mixed connective tissue disease
D Myofibrillar myopathy
E Myotonic dystrophy

Suggested Answer

B

Explanation / Comments

This man's symptoms and signs indicated an insidious onset of swallowing difficulty with more definite but asymmetric weakness of the wrist flexors. The description hints at mild facial weakness and possible early quadriceps weakness (a trip and difficulty rising from a kneeling position). The absence of pain or sensory features make radiculopathy or neuropathic weakness less likely. The pace of deterioration is slow, making motor neurone disease unlikely.

Myofibrillar myopathies are increasingly recognised, caused by a number of genes, and may cause late onset and distal weakness. Predominant distal arm weakness would be much less common than leg weakness in these heterogenous conditions. Myofibrillar myopathy is a possible but unlikely cause.

The late onset would not be compatible with a dystrophinopathy, and this X-linked disorder would not be inherited by a male through the paternal line.

Mixed connective tissue disease may be associated with a myositis, and this possibility is raised by his previous possible rheumatoid arthritis and Raynaud's phenomenon. The muscle weakness can be indolent and sometimes distal muscles are involved. However, there is no clinical evidence of active rheumatological disease, and the facial weakness would be unusual.

Myotonic dystrophy is common and may present at this age. It commonly causes distal weakness, but this would be unlikely in the absence of temporalis, masseter and/or sternomastoid weakness and wasting, or myotonia.

Inclusion body myositis (IBM) is the commonest muscle disease over the age of 50, and has an insidious onset which means the diagnosis is often made late. The commonest presentation includes quadriceps weakness, but the forearm flexors of the wrist and fingers are also commonly weak, and in some patients, as here, the presenting feature. The intrinsic hand muscles are relatively spared. Initial complaints explained to me include failure to hold onto a water skiing baton, open clothes pegs, or taking a wallet out of the pocket. Facial weakness and dysphagia are less common at presentation, but prominent in some patients. Most series of patients with IBM have noted a raised frequency of connective tissue disorders.

Confirmation of the diagnosis is by muscle biopsy. Some would use MRI in selected patients to show involvement of the quadriceps and other muscle in a suggestive pattern before biopsy. Two groups have recently identified an associated antibody in a proportion of IBM patients. No therapeutic agents are of proven use, but research is gathering pace.

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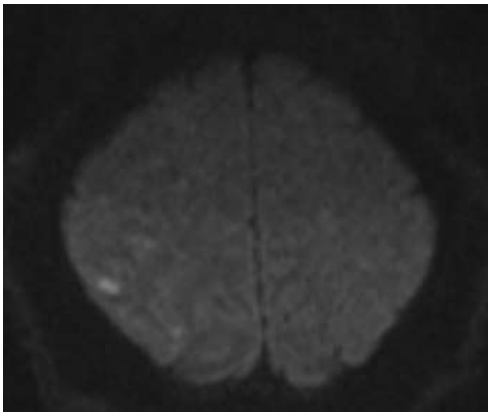
Question 3

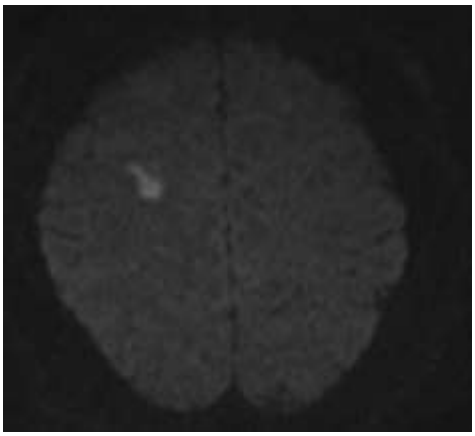
35 year old non-smoking male presented with a stroke causing a mild left hemiparesis. He had a past medical history of hypertension, impaired glucose tolerance, sickle cell trait (HbS-HbG Philadelphia) and obstructive sleep apnoea. On admission, he was taking Doxazosin MR 8mg, Lercandipine 10mg, Atenolol 25mg, Ramipril 10mg, Simvastatin 40mg. An aunt and cousin both had suffered strokes and type 2 diabetes.

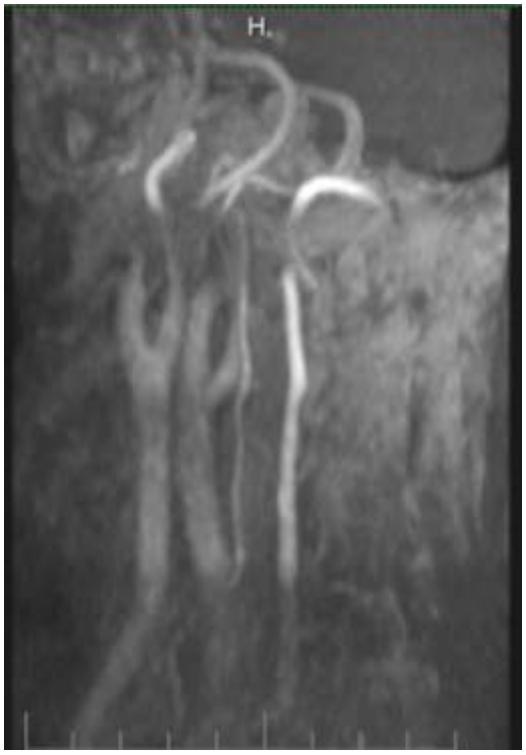
On examination, he weighed 128 kg with a BMI of 44.9. Pulse 53/min SR, BP 163/116, HS normal. There was mild left pronator drift, with grip in left hand 4+/5, reduced temp sensation in left arm and a left extensor plantar.

Investigations

- FBC normal, low MCV
- Electrolytes normal, K+ 3.2
- Cholesterol normal
- ESR, CRP, ANA normal
- Gluc 6.8, HbA1C 6.5%
- Factor V Leiden, ATIII normal
- ECG: sinus bradycardia
- Echo: normal
- MR imaging: see images







Q3 Answers

A 24h urine for metanephrines

B Arch aortogram

C Early morning serum cortisol

D Renal angiogram

E Serum aldosterone

Suggested Answer

E

Explanation / Comments

Results

•Renin <8 (recumbent 9.8 - 23.8 mU/L; erect 12.9 - 33.7 mU/L)

•Aldosterone 833 (recumbent 28 - 445)

Diagnosis - Conn's syndome (Primary hyperaldosteronism) resulting in resistant hypertension as another cause of his stroke (along with his obstructive sleep apnoea, type 2 diabetes and carotid dissection). A CT scan of his adrenals confirmed a 2.9 x 2.3cm oval solid mass arising from the lateral limb of the left adrenal.

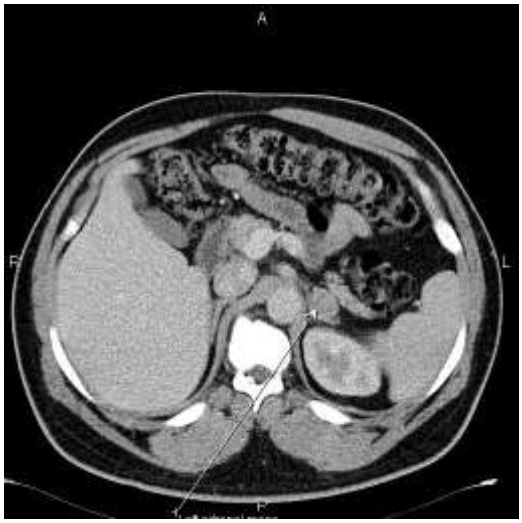
Serum sodium almost always > 140 mmol/l and serum potassium usually 3.5 - 4.0 mmol/l. However, potassium may be low normal with a high normal sodium in about 20% (personal experience of Prof Gareth Beevers in Birmingham who has seen 47 patients with Conn's in 33 years).

Rule of adrenal imaging -Don't do it unless there is an abnormality of cortisol or aldosterone (or the catecholesamines).

Take home messages

- Conn's syndrome is common, it's diagnosed late, it is "curable"
- Look at plasma Na^+ & K^+ "critically"
- In most cases a "random" renin & aldosterone will confirm or exclude the diagnosis

In this patient, he had been admitted in 2006 with uncontrolled hypertension and K+ 2.8, put on Sando K and GP advised to recheck, this did not happen!



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Acknowledgements / Consent (if applicable)

Patient consent for use of photo, I am grateful for Prof Gareth Beevers input on this case

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Question 4

A 26 year old female presented to the ED with an unusual spreading sensory sensation beginning in her knees, and increasing 'weakness' of her legs over 4 days. The sensory loss was described as 'wet' and had spread down her legs, then up the back before spreading onto the abdomen. She was assessed by the ST2 who felt her neurological examination was normal and she was discharged.

She returned the following day with increased weakness and was unable to walk steadily. Her gait was 'wobbly'. Bladder and bowel function were normal but she was having difficulty reaching the toilet in time. She could not stand unaided and tended to fall, especially when she closed her eyes. No reflexes were found in the lower limbs. Plantars were unresponsive. There were was a reduction in joint position sensation to the ankles. Her gait was felt to be functional.

She had a history of chronic severe renal colic and attended the ED regularly where a care plan for pain was lodged. There was no other medical history and there were no systemic symptoms. She did not smoke or drink much alcohol. She had just broken up with her long term partner 6 days prior to their wedding. She attended clubs on a regular basis and used 'balloons' at least twice per week. There was no family history of neurological disease.
Which test is most likely to confirm the diagnosis?

Q4 Answers

A. Lumbar puncture
B. MRI head and spine
C. Nerve conduction studies and EMG
D. Serum homocysteine
E. Serum vitamin B12

Suggested Answer

Answer: D

Explanation / Comments

Acute paraplegia has a wide differential diagnosis. Acute lower limb weakness is a presenting feature of Guillain Barré syndrome (GBS) which is so often missed. My experience of GBS is that about half of cases are seen and discharged at least once from A&E before the diagnosis is recognised. A high index of suspicion is needed early in the presentation when reflexes can be retained and sometimes disability is out of proportion to signs; an inappropriate discharge may be fatal in a rapidly progressive case.

Myopathy, neuromuscular junction disorders, electrolyte imbalance, and functional disorders are all included in the differential, but are less common. Acute myelopathy can mimic peripheral nerve disorders. The clue in this case is the sensory loss which began in a very unusual place and spread in a 'round the clock' manner. It also is much more complex ('wet') that would be described in the periphery. The imbalance, 'wobbly' gait, positive Romberg's test and joint position loss are all dorsal column symptoms.

The other clue is the social history. Balloons (for those that don't know) are balloons into which nitrous oxide is discharged and then inhaled. Also known as 'hippy crack', 'sweet air' or 'whippits' (the canisters drive Chantilly cream dispensers), balloons are used by more than 350 000 people per year, second only in users to cannabis in the UK. We all should be more streetwise and aware of this. This patient was also using nitrous oxide as part of her pain care plan and therefore had had chronic usage over a prolonged period.

Neurological vitamin B12 deficiency can be very acute and does not necessarily have any other associated haematological features of B12 deficiency such as a raised MCV; indeed in this case the serum vitamin B12 was normal (the lower limit of normal), but the homocysteine and methylmalonic acid were enormously raised demonstrating complete functional blockade of the B12 pathway. Nitrous oxide oxidises the cobalt atom in cobalamin from its 1+ to a 3+ valency, inactivating the active methylcobalamin as a cofactor in the methionine synthase pathway. Methionine is a precursor of s-adenosylmathionine which is essential in myelination and DNA synthesis and the inactivated methylcobalamin blocks its formation from homocysteine.

Immediate replacement of intramuscular B12 and folic acid resulted in some improvement. She was discouraged from using nitrous oxide for recreational purposes as should anyone else you know, especially your children!

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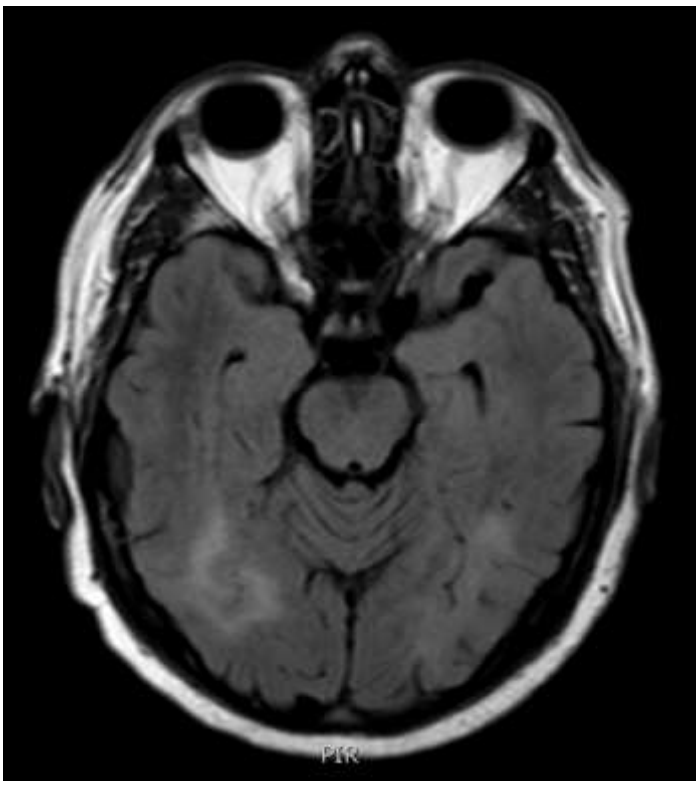
Question 5

A 59 year old male jewellery artist was admitted to hospital with a 6 month history of headache, progressive visual blurring and poor colour vision.

Past medical history included a diagnosis of interstitial pneumonitis made in 2008 when he presented with shortness of breath, skin lesions, arthralgia and a CT lung showing pulmonary fibrosis. A lung biopsy confirmed the diagnosis.



He was on Azathioprine 75 mg and prednisolone 10 mg for the above diagnosis.

Examination revealed no colour perception (black and white only) and visual acuities of hand movements only in both eyes. Normal anterior eye examination and fundoscopy. In addition he had a dysphasia with poor memory. General examination revealed purpuric skin lesions and telegangectasia.

Investigations:
ANA 1:1600
ESR 25 mm/hour
SSA (Ro) >50
HIV negative
CSF: acellular with a protein of 0.7 g/l, glucose: 3.1 (serum 4.7)
PCR: negative for JC virus & measles, no AAFB seen
Toxoplasma negative
Cryptococcal antigen (CRAG): negative
No oligoclonal bands
EEG intermittent non focal slow waves.
MRI: see image

What is the most likely diagnosis?

Q5 Answers

A Cerebral Vasculitis
B Mixed connective tissue disease
C PML
D SSPE
E TB

Suggested Answer

C

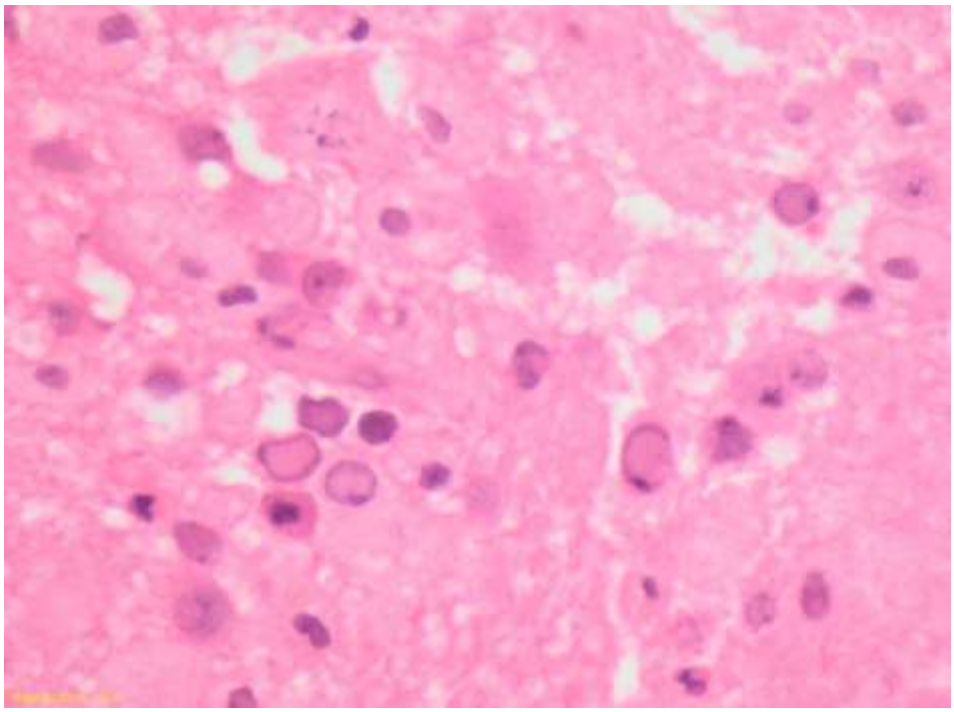
Explanation / Comments

The visual presentation of rapidly progressive loss of acuity together with cognitive problems in a patient on immunosuppression is very suggestive of PML, although it is rare in patients on azathioprine. PCR for JC virus is 76% sensitive but 96% specific.

SSPE is an important differential and gives very similar radiological findings but is typically accompanied by negative myoclonus and drop attacks. The patient is old for this diagnosis and we are not told he comes from an area of high prevalence of measles. The ocular findings are usually those of chorioretinitis which should have been picked up on ophthamic examination of this case.The absence of oligoclonal bands and negative PCR for measles RNA would be against the diagnosis. The EEG is often diagnostic.

The MRI appearance is not typical of vasculitis from either connective tissue disease or primary cerebral vascultis. The normal CSF makes TB less likely.

This patient went on to have a brain biopsy which showed hyaline inclusions in oligodendrocytes.



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Acknowledgements / Consent (if applicable)

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Question 6

You see a 67 year old man in Out-Patients. He had worked as a bank manager, taking early retirement at the age of 62 when he was offered an attractive retirement package.

Four years previously he began to have occasional episodes in which he stared blankly ahead and smacked his lips for seconds, with subsequent disorientation for several minutes. His wife also reported two occasions on which he had wet the bed at night over the past year, although they has slept separately for many years, so she could not provide any other history. Following one of these he had woken with a bitten tongue. He had not previously sought medical help with these symptoms.

Additionally, over the past three years his wife had noticed deterioration of his day to day memory, with difficulty recollecting recent events and conversations, and forthcoming events. He had become unreliable with the family accounts, which his wife now checked. Previously extrovert, he was now shy in company.

There was no past medical history of note, bar borderline hypertension for several years, treated with Bendroflumethiazide. He had never smoked, but drank 3 units/day. There was no relevant family history.

On cognitive assessment he scored 24/30 on the mini-mental state examination, 77/100 on the Addenbrooke's Cognitive Examination-Revised, with subdomain scores of 13/18 for attention and orientation, 16/26 for memory, 8/14 for fluency, 25/26 for language, 15/16 for visuospatial tasks.

There were no focal neurological signs.

Investigations:

- EEG: normal
- MR head: scattered high signal areas in the white matter, more numerous than expected for age. No signal change in the medial temporal lobes, but borderline atrophy on the right
- CSF: normal cell count, protein and glucose. CSF tau protein levels elevated, CSF abeta42 levels equivocally depressed
-

What is the most likely diagnosis?

Q6 Answers

A Alzheimer's disease
B Depression with dissociative attacks
C Limbic encephalitis
D Temporal lobe epilepsy with associated memory impairment
E Transient Epileptic Amnesia

Suggested answer

A

Explanation / Comments

The episodes described are typical of focal seizures with a temporal lobe origin, with probable secondary generalisation on two occasions at night.

The cognitive symptoms suggest a memory problem, primarily, with a possible disorder of 'executive function' (difficulties with accounts could be due to executive problems, memory problems or a more specific disorder of arithmetical ability). Social withdrawal is a non-specific symptom associated with cognitive decline, personality change or mood disturbance.

Bedside cognitive assessment confirms the presence of a cognitive disorder, particularly affecting memory and verbal fluency, in keeping with the history of a disorder of memory and executive function.

Thus the history and examination suggest a disorder involving epilepsy and cognitive impairment. The key question is how these are related. Does this patient have a neurodegenerative disorder causing secondary epilepsy, a primary epilepsy syndrome with secondary cognitive impairment, or a disorder which can give rise to both, such as limbic encephalitis? The relatively slow and insidious progression of cognitive decline in combination with relatively infrequent seizures makes a neurodegenerative cause the leading possibility, with Alzheimer's disease the leading candidate: Alzheimer's disease is a risk factor for epilepsy, and most commonly presents with memory disorder.

The normal EEG contributes little. The MRI scan shows non-specific white matter changes in keeping with the history of hypertension. The subtle medial temporal lobe atrophy would be in keeping with a neurodegenerative disorder, especially Alzheimer's disease – but is not definitive. The CSF changes are suggestive of Alzheimer's disease, in which the common profile is of elevated CSF tau protein concentration with decreased CSF Abeta42 concentration.

Among the diagnostic options on offer, depression can cause cognitive symptoms, including amnestic syndromes, but the description of the attacks is unlike dissociative attacks, and very typical for temporal lobe epilepsy. Transient Epileptic Amnesia presents with primarily amnestic seizures which is not the case here. Limbic encephalitis usually presents sub-acutely, over weeks, with severe memory impairment, temporal lobe seizures and behavioural symptoms, often accompanied by signal change in the medial temporal lobes on MRI. Memory impairment is a common complaint in people with epilepsy, especially when it involves the temporal lobes, but the scale of the impairment is beyond what one would normally feel comfortable in accepting for "just" epilepsy.

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None required
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Question 7

A 54 year old man presented with a 10 month history of gradually progressive weakness in his legs, later involving the arms, but without sensory symptoms or pain. He described no ptosis, diplopia, speech, swallowing or hearing problems; he occasionally had a dry mouth, and had lost some weight.

Past medical history: Epilepsy (levetiracetam, lamotrigine and sodium valproate) and CA colon treated with a hemicolectomy.

Ex smoker, 10-15 units of alcohol per week

On examination he had Grade 4-/5 power in shoulder abduction, elbow extension, hip flexion and knee extension bilaterally. Sluggish reflexes, down going plantars, sensory exam normal, cranial nerves normal.

Investigations

Routine blood tests: normal

Antibodies to Acetyl Choline receptor: negative.

Repetitive nerve stimulation (RNS): 28% decremental response at lower frequencies.

What would be the next most appropriate investigative step?

Q7 Answers

A Anti MuSK Antibodies

B Muscle biopsy

C Single fibre EMG

D Recheck Anti AChR abs

E RNS at higher frequencies

Suggested answer

E

Explanation / Comments

He had had LEMS which was clinically more likely than MG, but the neurophysiology was confounding, as the response was decremental rather than incremental. RNS in LEMS at lower frequencies can show a decremental response, but on higher frequency stimulation for longer periods of time, they should show an incremental response. The other neurophysiological option would be Post exercise CMAPs (PEF), which is less uncomfortable than high frequency RNS. Oh et al (2005) assessed both techniques in in 34 patients with LEMS. The sensitivity of using post-exercise CMAPS alone (taking any increment of ≥ 37% as positive) was 84%, for high frequency RNS alone (taking any increment of ≥ 47% as positive) was 97%, and using both, the sensitivity was 100%.

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Question 8

A 32 year old female had a twenty year history of type 1 diabetes, with mild retinopathy but no nephropathy and good diabetic control. She had had an episode of arrhythmia the year before, followed by a normal coronary angiogram. She was atopic, with hayfever and eczema. For the previous 3 months she had been dieting. Over the last six weeks she developed progressive sensory loss in the feet to ankles and mild proximal and distal weakness in all limbs with areflexia. She had features suggesting laser retinopathy which seemed extensive.

Nerve conduction studies showed a predominantly demyelinating neuropathy. CSF protein was raised.

What is the most likely diagnosis?

Q8 Answers

A B12 deficiency neuropathy
B Chronic inflammatory demyelinating polyradiculoneuropathy
C Diabetic neuropathy
D Mitochodrial disease with POLG1 mutation
E Refsum's disease

Suggested Answer

E

Explanation / Comments

Refsum's disease is a rare autosomal recessive disorder of fatty acid metabolism leading to a toxic accumulation of phytanic acid, and uncommonly, can present in adulthood. Symptoms can rapidly worsen during periods of weight loss. The mainstay of treatment is dietary avoidance of phytanic acid but plasma exchange is helpful for acute exacerbations.

This case was initially thought to be GBS but she did not improve with IVIG and atypical CIDP was considered. She improved significantly with plasma exchange. Her ophthalmological appearance was retinitis pigmentosa, she had cardiomyopathy, short fourth toes and the "eczema" was the scaly skin of Refsum's. Serum phytanic acid was grossly elevated.

References

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Acknowledgements / Consent (IF APPLICABLE)

Dr Rob Hadden (based on a similar case presented at the British Peripheral Nerve Society)

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Question 9

A 67 year old female presented with a 9 month history of intermittent, severe pain in the left lateral thigh. It had first occurred whilst on a cruise, and initially was not too bad; it would last for a few minutes, and there were no precipitants, although it often occurred in the morning. It settled for a few weeks, then returned, much more severe; she described agonising pain in the lateral thigh, lasting a minute or two, which would force her to stop whatever she was doing. It could occur several times within the first hour after getting up, then ease off, although could occur less commonly at any time of day. In between episodes she was well. Examination was normal, but she commented that when the pain was present, the lateral thigh felt numb.

What is the most likely diagnosis?

Q9 Answers

A: Functional pain syndrome

B: L2 radiculopathy
C: Malignant infiltration of the lumbar plexus
D: Meralgia parasthetica
E: Migrating sensory neuritis of Wartenberg

Suggested Answer

B

Explanation / Comments

This is an unusual (but real life) case. Although the distribution she described sounded precisely that of the lateral cutaneous nerve of the thigh, it is not typical of meralgia parasthetica, which is usually much less severe and less paroxysmal. It is not migrating anywhere, and the 9 month history and distribution is against a malignant plexus lesion. Beware of labelling unusual symptoms as functional just because you cannot think of a better explanation. I initially called this MP, but knew it was not right, and after a week of waking up in the middle of the night knowing I was wrong, capitulated and organised an MR lumbar spine to exclude an L2 radiculopathy (accompanied by a slightly embarrassed phone call to the patient). This revealed an unusually high prolapsed intervertebral disc, impinging upon the left L2 root. Although it initially (and rather unusually in my experience) responded to gabapentin, it recurred after a few weeks and was exceedingly troublesome, precipitating at least 2 episodes of reflex syncope such was the pain, and after much deliberation she underwent surgery, with an excellent, and persisting, good result. My own experience of PID (prolapsed intervertebral disc rather than pelvic inflammatory disease) is that first thing in the morning was often a particularly unpleasant time, and my neurosurgeon told me it was because overnight, the spine desiccates and shrinks down, enhancing pressure on the nerve root.

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Question 10

A 53 year old Caucasian woman presented with a 3-year history of progressive cognitive impairment, irritability, depression, unsteadiness and involuntary movements. She reported that her father died of dementia in his 70s but there was no other family history. She had no siblings.

A genetic test for the Huntington's disease CAG repeat expansion revealed two normal-length alleles.

Which of the following genetic tests is most likely to be diagnostic?

Q10 Answer

A: ATN1 gene: Dentato-rubro-pallido-luysian atrophy

B: HD gene: sequencing for point mutations

C: JPH3 gene: HD-like disorder 2

D: TBP gene: Spinocerebellar ataxia type 17

E: TITF2 gene: Benign hereditary chorea

Suggested Answer

D

Explanation / Comments

SCA17 is the commonest genetic cause of HD-like presentations in Caucasians among those presented, and a close HD mimic, especially with unsteadiness.

BHC is usually young-onset, doesn't cause progressive cognitive impairment and only rarely causes functional problems. HDL2 is essentially confined to Afro-Caribbean populations and unheard of in Caucasians. Sequencing of the HD gene is pointless - no pathogenic point mutations have been described. DRPLA is usually young onset and accompanied by seizures.

References

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Acknowledgements / Consent (if applicable)

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