

# Neurology

## Theme : Genetics - clinical abnormalities of limbs

- A. Bloom's syndrome
  - B. Cockayne's syndrome
  - C. Down's syndrome
  - D. Ehlers Danlos syndrome
  - E. Hunter's syndrome
  - F. Prader-Willi syndrome
  - G. Rubinstein-Taybi syndrome
  - H. Russell Silver syndrome
  - I. Sotos' syndrome
  - J. Williams syndrome
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In a child with a suspected genetic disorder and the following clinical abnormalities choose the single most likely diagnosis from the list of options.

1) Asymmetry of limbs and clinodactyly

H. Russell Silver syndrome

### Note:

In Russell-Silver syndrome skeletal features of this condition include short stature of pre-natal onset with asymmetry, most commonly of the limbs. There is a short inward curving to the 5th finger (clinodactyly).

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2) Sandal gap.

C. Down's syndrome

### Note:

In Down's syndrome 'Sandal gap' is found. This is wide gap between the first and second toes. Plantar creases between the first and second toes are usually deep and there is characteristic dermal ridge pattern. Simian creases are found in approximately 45% of babies with Down's syndrome. This is a single horizontal palmer crease. 85% of affected individuals have a distal positioning of the palmer axial tri-radius.

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3) Lax ligaments and joint hypermobility.

D. Ehlers Danlos syndrome

### Note:

In Ehlers Danlos syndrome there is joint and skeletal hyper-extensibility with poor wound healing. Blood vessels are fragile and this often manifests itself with recurrent bruising. Affected individuals tend to have characteristic facial features including epicanthic folds, blue sclerae and a narrow mandible.

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### Comments:

Examination of the hand and feet are important in the assessment of a dysmorphic infant.

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## Theme : Neurological investigation

- A. Angiography
  - B. CT head scan
  - C. ECG
  - D. EEG
  - E. Lumbar puncture
  - F. MRI scan
  - G. No investigation required
  - H. Skeletal survey
  - I. Skull x-ray
  - J. Tensilon test
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**Select the most appropriate investigation for the following cases**

1) A 6 month old baby with microcephaly is brought to A&E. His mother gives a history of the baby flexing forwards whilst sat in her highchair. **D. EEG**

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2) A 6 year old boy with a previous history of convulsions and abnormal EEG's presents with tingling sensations in his right thumb which precedes rapid jerking of his right arm. **B. CT head scan**

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3) A 2 year old boy with an otitis media is brought to casualty. His father reports that the child has had a fit. Temperature recorded is 38.8°C. **G. No investigation required**

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**Comments:**

Item 1 relates to a baby with infantile spasms. Brief symmetrical contractions of the neck, trunk and extremities. It is related to pre-natal, peri-natal and post-natal causes for example hypoxic ischaemic encephalopathy, congenital infections and metabolic disease. The baby requires an EEG and this will show hypsarrhythmia, a chaotic high amplitude slow wave and spike abnormality. Item 2 describes a boy with focal seizures, previous EEG's have been abnormal. A CT scan is indicated to exclude underlying space occupying lesions. The treatment of choice would be Carbamazepine. Febrile convulsions are very common they manifest as generalised tonic clonic seizures and occur with rapidly rising temperatures. 50% of children having had one febrile convulsion are likely to have a second. No investigations are required.

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**Select the most appropriate investigation for the following cases**

1) A 5 year old boy presents with screaming at night usually between midnight and 2am.

G. No investigation required

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2) A 3 month old with a coryzal illness presents to surgery for his first immunisations. He has bruising to both cheeks and mother reports that it is due to rolling off the settee.

H. Skeletal survey

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3) A 3 year old boy is brought to hospital for investigations with a history that he is progressively getting more tired by the end of the day and an inability to swallow.

J. Tensilon test

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**Comments:**

Item 1 describes a child having night terrors. These are very common (approximately 3% of all children). Typically a child between the ages of 5 and 7 will awake between the age of 12am and 2am screaming. They appear frightened, they may have a tachycardia and dilated pupils. Children tend to fall back to sleep with total amnesia of the event the following morning. Item 2 describes a baby having sustained physical, non-accidental injury. This diagnosis must be suspected if medical findings are unexplained or the history offered is inconsistent. Also if the child suggests that the injury has been caused by an adult. Characteristic injuries include bruises as in Item 3 and further investigations including blood tests, skeletal survey, plus or minus a CT head scan must be considered. The last question describes a child with progressive weakness, hypotonia and an inability to suck and swallow. Myasthenia Gravis must be considered, this is a defect in the neuromuscular junction with acetyl choline receptor antibodies. A Tensilon test may show an increase in muscle strength.

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**Theme : Side effects of Anti-Convulsants**

- A. Carbamazepine
  - B. Clonazepam
  - C. Ethosuximide
  - D. Gabapentin
  - E. Lamotrigine
  - F. Phenobarbitone
  - G. Phenytoin
  - H. Sodium Valproate
  - I. Topiramate
  - J. Vigabatrin
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**Select one of the drugs from the list of options that corresponds with the listed side effect:**

1) Increased appetite resulting in weight gain.

H. Sodium Valproate

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2) Visual field defects.

J. Vigabatrin

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3) Is associated with hyperammonaemia.

H. Sodium Valproate

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**Comments:**

Sodium Valproate has many side-effects including gastric irritation and nausea as well as Hyperammonaemia (interferes with the urea cycle, reducing Carnitine concentrations and has been associated with hyperammonaenic encephalopathy), increased appetite and weight gain and transient hair loss.

Vigabatrin is used for treatment of partial epilepsy with or without secondary generalisation, usually in combination with other anti-epileptic drugs although can be used as monotherapy in the management of infantile spasms. Side-effects include drowsiness and confusion and visual field defects.

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**Select one of the drugs from the list of options that corresponds with the listed side effect:**

1) Needs to be withdrawn if a rash develops.

E. Lamotrigine

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2) Coarse facial features, hirsutism and gingival hypertrophy.

G. Phenytoin

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3) Anorexia leading to weight loss.

## I. Topiramate

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### **Comments:**

Lamotrigine is an anti-convulsant used for treatment of partial seizures as well as generalised seizures. It is also used for myoclonic seizures, atypical absences and Lennox-Gastaut syndrome. Lamotrigine may cause a serious skin rash in children and consideration must be given to withdrawal of the drug should the rash develop.

Phenytoin is effective in the treatment of tonic-clonic seizures and partial seizures. It may cause facies, acne, hirsutism and gingival hyperplasia.

Topiramate can be given as adjunctive treatment for partial seizures with or with secondary generalisation. One of the main side-effects is anorexia leading to weight loss.

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