Epilepsy mimics
George Cheyne 1733
The English Malady

“dropping down suddenly, as if shot; but sometimes with violent Convulsions and Throws in the Belly, Breast and Limbs, beating and tearing themselves, clinching their Fists, biting their Tongues, grinding their Teeth and foaming at the Mouth, with a small Trembling, unequal and sometimes interrupted Pulse, and an involuntary secretion of all natural Discharges.”
George Cheyne 1671-1743
Staring and dreamy states could also be manifestations of epilepsy

- Tissot 1770: “petit mal” as well as “grand mal” seizures in a 14 year old girl
- Heberden 1802: “half fainting, joined perhaps with forgetfulness or delirium for a few minutes”
- All described in India 1000 BC “Ayuverda”
Why bother? So last century?

- Increasingly recognised that there is an overlap in the underlying pathophysiology of epilepsy, movement disorders and migraine
- May respond to the same treatments
- Does making a correct diagnosis matter in the real world?
Common areas of difficulty in the diagnosis of seizures

- Babies that jump around
- Convulsive syncope
- Psychogenic non-epileptic seizures
What is the secret of diagnosis?

You must remember this
A kiss is just a kiss, a sigh is just a sigh. The fundamental things apply
As time goes by.

Porter C. (1931)
History

LORD KYSANT, WITH HIS SON-IN-LAW, THE EARL OF COVENTRY, LEAVES FOR MR. JUSTICE WRIGHT'S SUMMING UP IN THE ROYAL MAIL STEAMSHIP CASE
Importance of the history

- Taking a careful history may enable you to make a diagnosis before any tests are done.
- It communicates to the child and parents you are interested in them and are careful.
- May need explanation: some parents and children perceive it more as a cross examination.
History = Systematic

- What was the child doing when the event occurred?
- What was the first sign of something going wrong?
- How did the event evolve? (step be step)
- What was the child like afterwards? How long before they were back to normal?
Don’t allow every event the child has ever had to be combined into a single distorted and confused tale.
- Go to the most recent well remembered typical event
- Once what happened at that time is established, go to other events to see if they are all the same or not
Moving babies and infants

- Babies and infants who
- jerk, twist, shake, shudder, twitch, wheel, fling, roll
- and make people nervous
2 week old baby

“Generalized tonic-clonic seizures”

No response to phenobarbitone, phenytoin or clonazepam
Benign Neonatal Sleep Myoclonus

- Repetitive myoclonus only during sleep
- Quiet sleep more than REM sleep
- Onset often in week 1, increasing intensity until week 3
- Generally stop by 6 months, 97% by 12 months
- Jerks: unilateral, bilateral, repetitive, violent
- May appear as short clusters that shift sides.
Benign Neonatal Sleep Myoclonus

- In some infants, repeated multifocal jerks continue for 30 minutes or more
- Clinical exam & EEG normal
- No response to anti-epileptic drugs (may be made worse by them)
- May be a positive FH
- *Well baby having multiple prolonged daily seizures* – think of BNSM
Shuddering attacks

- Episodes last usually a few seconds
- Combination body stiffening and trembling
- “Water poured down back”
- Onset 3-6 months, after 3 years
- Spells variable frequency, >100/day
- Provoked excitement, anger, fear, frustration,
Shuddering

- Vanasse et al (1976)
- Postulated that early manifestation of essential tremor
- Not confirmed over time
- High incidence of tics in the families in the original paper
Benign Myoclonus of Early Infancy

- Fejerman and Lombroso 1976, 1977
- myoclonus
- spasms and brief tonic contractions,
- shuddering,
- atonia or negative myoclonus,
- combinations of these movements
Benign Myoclonus of Early Infancy

- Tendency to come in clusters
- Can mimic infantile spasms
- Most often awake
- Examination and EEG normal
- Hard to get a firm grasp on this because of variability of presentations
Benign Polymorphous MDs of Infancy
Benign Polymorphous MDs of Infancy

Movements can be abruptly stopped “before their time” by surprise tactics
e.g. a sudden unexpected tickle
- Neonate
- Attacks precipitated by wiping perineum
- Father had similar episodes
Familial Rectal Pain (Hayden & Grossman 1959)

- OMIM: %167400: Pain, Submandibular, Ocular and Rectal, with Flushing
- Paroxysmal Extreme Pain Disorder (Fertleman & Ferrie 2005)
- JBP Stephenson: PAIN: Paroxysmal Autonomic Intolerable Neuralgia
Paroxysmal Extreme Pain Disorder

- Onset neonatal period, perhaps in utero
- Attacks of excruciating pain that can affect various parts of the body including rectum, genitalia, face & limbs
- Harlequin colour change, pupillary abnormalities
- Flushing (particularly around buttocks), oedema
- Non-epileptic tonic seizures during attacks of pain
- May be associated with cardiac asystole
Precipitation pain & attacks

- Bowel motions
- Feeding
- Loud noises
- Choking
- Sneezing
- Being put in a hot bath
- Touching
- Spontaneous
Paroxysmal Extreme Pain Disorder

- Worst pain ever experienced, 100/10
- Rectum spread to whole body then collapse
- Jaw pain when eat
- Eye pain: wind on face “red hot needle”
Paroxysmal Extreme Pain Disorder

- Autosomal dominant
- Mutation SCN9A: encodes voltage gated Na channel Nav 1.7 (Fertleman et al 2005)
- Erythromelalgia: mutations in the same gene
- Expressed in peripheral sensory & autonomic neurons
- ? Hyperexcitability and repetitive firing of sensory neurons
Paroxysmal Extreme Pain Disorder

Treatment

- Immersion in cold water
- May respond to carbamazepine
- ? Mexiletine
- Atropine/pacing
- Inhalation nitrous oxide
Imai et al 2015

“Short-lasting unilateral neuralgiform headache attacks with ipsilateral facial flushing is a new variant of paroxysmal extreme pain disorder.”
5-year-old girl

- recurrent, unilateral temporal headaches
- lasting 20–90 s
- associated with ipsilateral conjunctival injection, lacrimation, nasal congestion, rhinorrhea, and facial flushing.
- Headaches often triggered by hitting her head or body, taking a bath, experiencing a temperature change, or sleeping.
- 10–20 times per day.
Flushing of (A) the face and (B) arm after a right-sided severe headache attack in Patient 1.
Dominant FH
heterozygous missense mutation in SCN9A
carbamazepine 100 mg/day resulted in complete disappearance of the attacks
Familial episodic pain syndrome

- begins in infancy and triggered by fasting or fatigue
- other contributing factors include illness, cold temperature and physical exertion
- caused by an autosomal-dominant mutation in the gene for TRPA1
12 months old
Progressive loss of skills over 3 months
Given treatment
Suddenly started jerking
Vitamin B12 (cobalamin) deficiency

- Combination myoclonus and tremor
- Often misdiagnosed as seizures
- Seizures can also occur
- MD most commonly appears after treatment
- Purely breast fed infant
- Mother unrecognised B12 deficiency (vegan or pernicious anaemia)
- Macrocytosis but may not be anaemic
Self Stimulation Episodes

- “infant masturbation”
- may mimic epilepsy, movement disorder, intra-abdominal emergency
- usually female infants using thigh friction
- Still 1915
- 22/25 were less than 2 when started
- ? vulval irritation
- can occur many times per day
12 year old boy
- ? Degenerative disease
- Deteriorating school performance
- Frequent seizures unresponsive to AEDs
- “It would be good to see an event”
Cataplexy and similar episodes

- Narcolepsy/cataplexy
- Niemann-Pick type C disease
- Coffin-Lowry syndrome
- Norrie Disease (Vossler 1996)
- Prader Willi syndrome
- Pontomedullary/hypothalamic structural lesions
Vasovagal episodes common: sudden loss of adequate cerebral perfusion
Jerking movements are common in syncope

- Recreational self-induced syncope in adolescents (Jeong et al. ESA 2015)
- Hyperventilation
- followed by sudden standing, chest compression or Valsalva manoeuvre
- 266 videos You Tube
- 44.2% had convulsive movements
Stretch syncope
Pelekanos et al. 1990

- 6 adolescents with syncope induced by stretching with the neck hyperextended.
- mechanism is not simply Valsalva
- may also involve vertebral artery compression coupled with a familial tendency to faint.
Evans et al. 2009

- 1525 patients with syncope
- 111 hair-grooming trigger
- 78% were girls
- girls experienced syncope more during hair combing and brushing
- boys during hair cutting
- benign form of neurocardiogenic reflex syncope
Prolonged Q-T Syndrome

Bailey 1996

“These patients are often misdiagnosed even by experienced neurologists as suffering from epilepsy.

For some this may be a fatal diagnostic error.”
Genes associated with prolonged Q-T syndrome

*KCNQ1* (locus name LQT1), *KCNH2* (LQT2) and *SCN5A* (LQT3) are the most common. Other, less frequently involved genes are *ANK2* (LQT4), *KCNE1* (LQT5), *KCNE2* (LQT6), *KCNJ2* (LQT7), *CACNA1C* (LQT8), *CAV3* (LQT9), *SCN4B* (LQT10), *AKAP9* (LQT11), *SNTA1* (LQT12), *KCNJ5* (LQT13), *CALM1* (LQT14), and *CALM2* (LQT15)
Breath holding attacks provoking epileptic seizures

Sometimes the seizures are prolonged
“Less well known is the situation in which a syncope - that is, an anoxic seizure - provokes a true epileptic seizure.

This combination is called an (AES) and was first described as such by one of us in 1983.”

27 children seen over 30 years in Glasgow
Anoxic epileptic seizures

- Prophylactic sodium valproate or carbamazepine abolished epileptic component in 5 of 7
- Syncopes continued unchanged.
- Syncopes far more common than Sz
- In 3 children in whom ratio could be calculated, about 7–8% of syncopes developed into AES
10 year old boy

- Sudden loss of consciousness and falling after unexpected physical shock
- tripping on tree root,
- being poked in the ribs from behind by another child
- no problems in soccer as anticipating a knock
RAS
reflex asystole from surprising pain
Sudden unexpected pain, fright, surprise

Convulsive syncope
Vasoccardiac anoxic seizures
Fig. 9.4. Duration of ocular compression induced asystole in 39 children who had a clinical diagnosis of vagocardiac reflex anoxic seizures. (The two adults in the series did not have ocular compression.)
“I write as the day unfolds over the changing waters of Loch Indaal and the light in the west tells of the golden eagle soaring close by Lossit bay quartering the salt-sprayed machair.”
Ronnie MacKeith (1975)
Or is a children’s physician just naturally nice?

- Mild hypomania
- Obsessional traits
- Insecurity
- Masochism

“Embrace and make good use of your neurotic traits.”