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OPINION FORUM

Neutralising Neurophobia

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Introduction

Medical students are being trapped in the void between basic sciences and clinical facts. Of greater concern is that the chasm is widening with the progressive decline of the basic sciences, which require years to master. Meanwhile we expect our doctors to disperse quickly and start righting the wrongs. Teaching laboratories are becoming more sophisticated in order for students to be able to intubate and put up CVP (central venous pressure) lines unsupervised. Cardiac murmurs are digitalised, recorded and replayed. Mitral valve prolapse is an electrical oscillation of red lights and not the whooping of a white dove. The experience is made as real as possible and becomes ever more surreal. Medicine becomes performance based and frozen. The patella hammer rests on top of the book.

Simulating reality and understanding reality are entirely different. We watch for the smoothness and flow of reflexes during student examinations. Yet one third of normal children will have brisk reflexes, another third will have normal reflexes, and the final third will have depressed or entirely absent reflexes. Therefore a paediatrician who may be a master clinician will be unable to elicit reflexes in at least one third of his or her patients. A reflex is neither a sensitive nor a specific sign and the level of its execution should not be used as the main marker of clinical competence in the neurological examination. Of far greater clinical value is whether or not the reflexes are symmetrical and spread uninhibited. The correct implementation of theory limits the burden of expectation, and makes way for reasonable clinical practice. The fear contained within the academic hospital walls dissipates, and self-reliance replaces the name tags. This is freedom, and now you may call a university your "alma mater", which means "nourishing or kind mother" in Latin.

Basic sciences and clinical teaching cannot be taught separately. They are one and the same and must be valued as such. With universities under ever-increasing pressure to perform in accreditation ratings, it is time to reconsider what it truly means to leave a legacy. Surely our goal must be to put the man above the methodology? The man in front of you. The one who was sent through your door. This personal crusade of the teacher imprints onto the heart and mind of the learner. A young doctor who has a reference point will tend to listen more and speak less. Teaching has become instrumental in creating empathy. Neurophobia is the fear of the neural sciences and clinical neurology that is due to the students' inability to apply their knowledge of basic sciences to clinical situations.[1] Central circuitry becomes baffling, or impossible to understand, and the individual clings to peripheral reflexes for clarity, at the expense of the cranial nerves. The purpose of this article is to demonstrate by means of a case presentation how the complexity of the brain can be unravelled by gently laying down the patella hammer, and remembering the lectures.

History

A three-year-old boy presented with a history of generalised epilepsy. Seizure onset was during the neonatal period and remarkably, the perinatal history was normal. He was the mother's first baby and was born via normal vaginal delivery at 42 weeks' gestational age. The birth weight was 2.57 kg and the head circumference was 33 cm, both within normal limits. The Apgar Scores were 9/10 and 10/10 respectively and he never developed neonatal jaundice. At the time of presentation, the immunizations were up to date and there was no evidence on the antenatal card of any maternal infections during pregnancy.

Development

He had global and marked developmental delay. His cognition and speech in particular were severely delayed. He made simple vocalisations, cried persistently and was inconsolable. He was not smiling or casting (able to let go of an object). He could only sit in the tripod position and was unable to roll over from the prone position or bear weight. Although he had previously been diagnosed with cerebral palsy, his head circumference (50.5 cm) was normal and not crossing centiles. The mother thought that the child could see but was unsure whether or not he could hear.

Clinical examination

The striking feature was marked opacification of both eyes. The pupillary reflexes were intact and the red reflex was present bilaterally which indicated that the pathology was not due to cataracts. He was also able to see and track objects while moving his eyes in all directions. This localised the pathology to the cornea and on closer examination bilateral corneal scarring could be observed. The occurrence of corneal opacification is very unusual in paediatrics. According to the mother, she had noticed the corneal lesions already in the first few weeks after birth. This lead to the suspicion of either an underlying metabolic condition with recurrent episodes of hypoglycaemia and corneal scarring or a congenital TORCH infection as possible aetiologies.

The mother also reported that the child was unable to close his left eye during sleep, and that there was persistent drooling from the left corner of the mouth. There was indeed a lower motor neuron (LMN) palsy affecting the left side of the face. It was an isolated facial palsy as there was no corresponding limb weakness to suggest an intrauterine stroke. The power and tone in the limbs was normal and he could move all limbs symmetrically. The reflexes were brisk and with spread, indicating a supratentorial lesion and there was bilateral ankle clonus. There was also truncal hypotonia as well as truncal ataxia which suggested cerebellar involvement. There was no associated movement disorder or signs of basal ganglia pathology. The child was failing to thrive and had significant difficulty with feeding and swallowing. He was not dysmorphic and there were no signs of a neurocutaneous disorder. On general examination, there was no hepatosplenomegaly to suggest infection or metabolic storage. (Herpes simplex virus type 1 (HSV-1), which is also associated with cold sores, is acquired during early childhood and is responsible for the majority of corneal blindness in the developing world. Reactivation may result in herpes simplex keratitis and corneal scarring. However, a single eye is usually affected.[2]) No other abnormalities were detected on systemic examination.

Discussion

In this case, if theory were to dominate clinical examination, the clinician would approach the topic of congenital corneal opacification (CCO).Primary causes are hereditary or developmental abnormalities of the cornea, such as corneal dystrophies or glaucomatous CCO.Secondary causes would include infections (HSV-1 keratitis), iatrogenic injury (amniocentesis or forceps delivery) and metabolic disease.[3] As metabolic causes rarely occur at birth or within the first four weeks post-partum, and herpes simplex keratitis only develops in early childhood, the aetiology of the corneal opacification in this patient remained unclear.

This child actually presented with a unique cause of congenital corneal opacification. Resolution to the clinical problem lay firmly rooted in the basic sciences and could be easily obtained by means of a piece of cotton wool. The attending doctors had correctly ascertained that the patient had a LMN facial palsy on the left side together with feeding and swallowing difficulties and probable hearing impairment. However, certain questions remained unanswered. Firstly, they did not understand why the child was unable to swallow in the presence of a functioning gag reflex. The gag reflex relies on cranial nerve IX providing sensation to the back of the tongue and throat and vagus eliciting the motor response. As cranial nerves IX and X form a reflex arc, they generally do not function independently. Secondly, they could not establish a link between the corneal opacification and cranial nerve deficits. Lastly, they were unable to account for the cerebellar pathology.

"Wisely and slow - they stumble that run fast."

(William Shakespeare)

The reason why the child could not swallow in the presence of an intact gag reflex is because the glossopharyngeal nerve also innervates the stylopharyngeus muscle. The function of this tiny muscle is to elevate the larynx and pull it forward during swallowing so that food is passed to the oesophagus while bypassing the vocal cords.[4] This minor adjustment to the mechanics of the head and neck is critical in the prevention of aspiration. The fact that the child could not swallow but was able to gag revealed the presence of a brainstem lesion that selectively involved the medulla.

The doctors had seen the child blink on the right side but had failed to test for the corneal reflex. Although the absence of the blink reflex on the left could be explained by the LMN facial palsy, they had incorrectly assumed that because it was working on the right, this implied that all of the components of the reflex were intact. The sensory afferent of this reflex arc is supplied by the trigeminal nerve (corneal sensation) and the motor efferent to the orbicularis oculi muscle derives from the facial nerve. After stimulating the right cornea with cotton wool it was noted that the corneal reflex was absent in the right eye. The child was still able to blink with the right eye due to the dual innervation which the nuclei of cranial nerve VII receive from the unilateral as well as the contralateral cerebral hemisphere. The upper motor neuron (UMN) component provides voluntary control to the act of blinking, which is independent of the blink reflex. Thus the child could blink if he wanted to but would not shut his eye in response to corneal irritation or stimulation. This subtle discrepancy signified isolated fallout of cranial nerve V and placed the lesion specifically in the pons.

The link between the multiple cranial nerve fallout and the cerebellar signs can be found in the origins of embryology. The pons, medulla and cerebellum are formed together and constitute the hindbrain. The facial palsy, loss of corneal sensation with corneal scarring and hearing impairment represented the pons. The swallowing and speech difficulties with failure to thrive represented the medulla. The truncal hypotonia, ataxia and delayed motor milestones represented the cerebellum. By testing each cranial nerve thoroughly, an entire symptom complex emerged which revealed a distinctive and rare hindbrain malformation.

Pontine tegmental CAP dysplasia

The MRI findings were suggestive of a condition called pontine tegmental cap dysplasia (PTCD).There was flattening of the ventral portion of the pons and a "cap-like" projection extending from the rear of the pons posteriorly in to the fourth ventricle. This dorsal projection is in fact ectopic white matter, situated on the dorsal surface of the pons and is an aberrant extension of the pontine tegmentum. There was also hypoplasia of the cerebellar vermis and cerebellar peduncles, the latter giving the characteristic "molar tooth" appearance.[5] The condition was originally described by Barth et al. in 2007 and since then just over twenty cases have been described in the literature. Small case series have been described by Barth et al. (2007), Jissendi-Tchofo et al. (2009) and Briguglio et al. (2011), and none of these patients had a family history of the disorder. The occurrence therefore seems to be sporadic.[6] The malformation primarily involves the brainstem and hindbrain and its MRI appearance is distinctive.

Cranial nerve abnormalities involving the pons and medulla are prevalent, such as sensorineural deafness and difficulties with feeding and swallowing. As the fifth and seventh cranial nerves lie in close proximity to each other, they can be mutually affected, resulting in impaired corneal sensation and facial paralysis. Defective corneal sensation leads to malfunction of the blink reflex and corneal scarring together with impaired vision. Truncal hypotonia and cerebellar ataxia result from involvement of the cerebellar peduncles and vermis and less frequently, gaze palsies or oculomotor apraxia occur due to involvement of the abducens nerve.[7] Invariably significant cognitive delay is present which may be accompanied by epilepsy.[8] Pyramidal signs such as brisk reflexes and ankle clonus may reflect supratentorial involvement.

Conclusion

A misconception in neurology is that one has to wait for "dead man's boots". The great British neurologist, Professor Marsden, competed for and won his chair in neurology at the age of thirty four.[9] His distinction lay not in that he had won but in that he had "dared". The reason why very few know about the brain is because very few have ventured towards it. Not because they have failed. The brain is not infinite. It is not even a reflection of God but a gift. It does not require us to lie prostrate with our eyes facing the ground. The tragedy in neuroscience is that most wait for a cipher of the code to be handed down and barely hold on to it, let alone share it with others.

Adopt the cranial nerves as your introduction to neurology. Drooling and spasticity of the limbs signify an upper motor neuron lesion within the cerebral cortex and a pseudo-bulbar palsy. Drooling and flaccidity of the limbs implies the presence of a lower motor neuron lesion involving the brainstem and a bulbar palsy. Cranial nerve knowledge is essential and prevents the student from sifting through irrelevant clinical facts in order to conjure up a diagnosis. Our minds are phylogenetically wired to respond to gestures – the professor's gestures, the patient's gestures, the student's gestures.[10] The bedside must supersede didactic lecturing and become the inner sanctum of mime and cognizance.

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