Case 6 – Answer Key

Case A: You are working at a Pediatric Hospital.

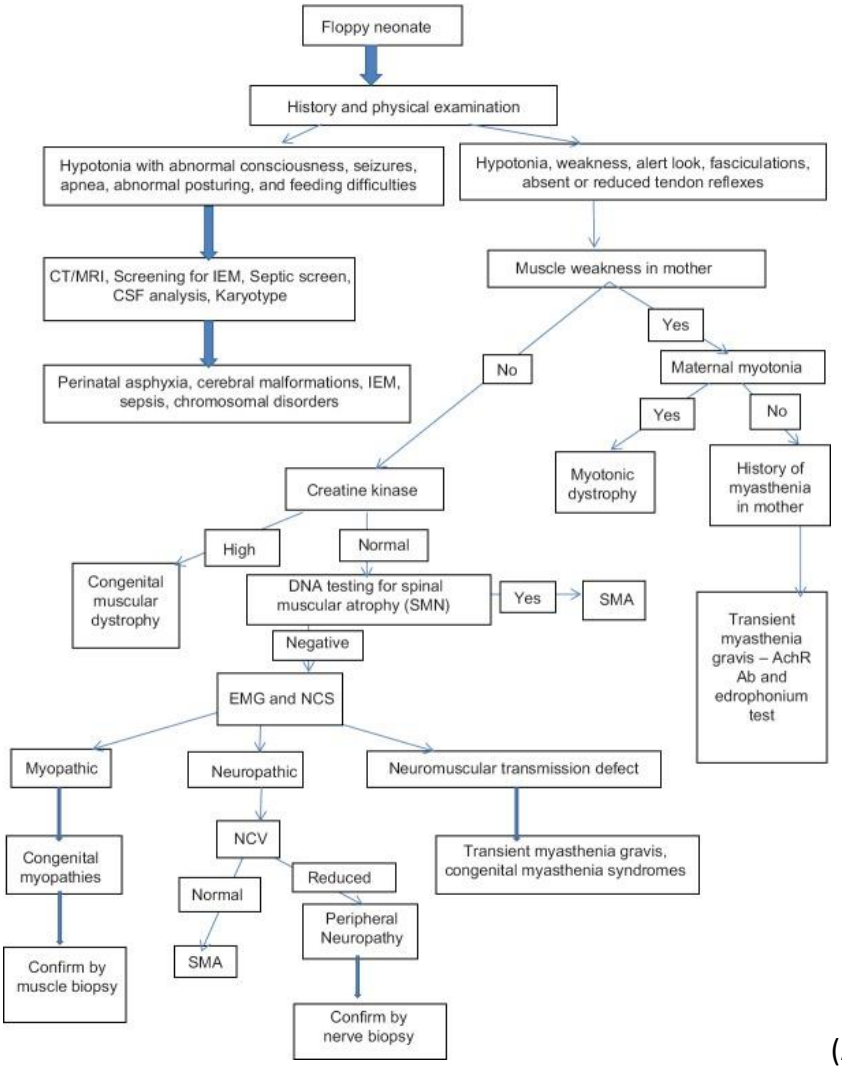
You are the pediatric physiatrist in charge for the day. You receive a call to assess a "floppy" neonate who is 1 mo.

1. **What is your differential diagnosis?**

Potential answers: CP, SCI/Spina bifida, SMA, peripheral neuropathy, congenital myasthenic syndromes, botulism, myasthenia gravis, myopathies, myotonic dystrophy, chromosomal disorders, metabolic diseases Acute presentation of hypotonia: sepsis, inflicted injury (accidental or non-accidental), congestive heart failure, electrolyte abnormality, GBS, Poliomyelitis, medication side effect.

REFERENCE : https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4862282/

2. **What investigations would you like to order?**



(Ahmed, 2016)

REFERENCE : https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4862282/

Case 6 – Answer Key

At this point, ***cerebral palsy*** is your main working diagnosis.

The parents are asking you how cerebral palsy is diagnosed.

3. **What do you want to tell the parents?**

CP is a CLINICAL DIAGNOSIS. There is no specific diagnostic test. Diagnosed by history, physical exam, serial clinical assessments, and neuroimaging.

Potential answers: CP describes a group of permanent disorders of the development of movement and posture, causing activity limitation, that are attributed to non-progressive disturbances that occurred in the developing fetal or infant brain. The motor disturbances of CP are often accompanied by disturbances of sensation, perception, cognition, communication, behaviour, epilepsy, and secondary MSK problems.

Brain MRI findings of periventricular leukomalacia/changes, brain maldevelopment, HIE, perinatal stroke.

Signs and symptoms suggestive of the diagnosis of CP:

- Abnormalities in muscle tone and posture (i.e. poor head control, asymmetric limb movement, hand preference, fisting)
- Delays in motor milestones, regression of skills
- Abnormal developmental reflexes
- Presence of associated conditions such feeding difficulties, growth failure, or epilepsy
- Parental concerns

REFERENCE : (Rosenbaum et al., 2007)

4. The child is now 6 years old. You see him for a concern of hypersialorrhea.

   What would you suggest?

Potential answers: behavioural (biofeedback techniques), orosensory/motor intervention, positioning, suction, hydration, medication (anticholinergic agents atropine drops-scopo-glycopyrolate-benztropin-trihexyphenidyl hydrochloride, botulinum toxin), surgery.

REFERENCE : https://www.aacpdm.org/publications/care-pathways/sialorrhea-in-cerebral-palsy

5. You are still following this patient. Now he is 17yo. You want to transition him to adult (ie. non-pediatric) services and talk to him about important topics as an adult living with cerebral palsy.

What are some important things you want to tell him and his parents?

Potential answers: adult clinic, transition, school, work, driving, financial, independent living, sexuality, relationship, mobility, social security, life skills

Live add-on question: should you have started the transition to adulthood earlier?

REFERENCE : Canadian Association of Pediatric Health Centres (CAPHC), National Transitions Community of Practice (2016). A Guideline for Transition from Paediatric to Adult Health Care for Youth with Special Health Care Needs: A National Approach http://ken.caphc.org/xwiki/bin/view/Transitioning+from+Paediatric+to+Adult+Care/A+Guideline+for +Transition+from+Paediatric+to+Adult+Care

Case B

You are seeing another "floppy" infant in your clinic. You are this time thinking it looks more like SMA.

6. **The parents are asking what spinal muscular atrophy is and what are the symptoms. What will you say?**

Potential answers: an autosomal recessive disorder with the absence of SMN1 gene (5q chromosome). Resulting in motor neuron disease.

Potential answers: bulbar (dysphagia, dysarthria, dysphonia, tongue atrophy, tongue fasciculations), appendicular (symmetrical proximal lower>upper extremity, delayed milestones, difficulty with stairs, falling, hypotonia, absence DTR, fasciculations, hand tremor, contractures), trunk (hyperlordosis, scoliosis, pulmonary restrictions (impaired cough, secretions, hypoventilation) and complications)

REFERENCE : https://www.ncbi.nlm.nih.gov/books/NBK560687/

7. **The parents heard on the radio that a cure might be available and would like to hear more about it. They can't remember the name of the treatment. What can you tell them?**

Potential answers: Nusinersen (ASO SMN2), Risdiplam (small molecule SMN2), Onasemnogene abeparvovec (gene therapy SMN1)

Live add-on question: What do you think about the price of those treatments? Should they be given for life? Could we give more than one treatment?

REFERENCE : https://www.ncbi.nlm.nih.gov/books/NBK560687/