Patient Journey Pitt-Hopkins Syndrome

Note: Information to parents and contact with support groups if desired!

… diagnosis can take many years …

Note: ideally we will find a treatment one day for the breathing anomalies and other ANS dysfunction

1 … usually normal pregnancy …

2

3

Note: Diagnosis explained many parents are given a print out of a medical paper and sent on their way!

4

5 … continuing need for physiotherapy and other therapies. …

1. & 2. Pre-Diagnosis: Usually normal pregnancy. Some ultrasounds may show something, some may have initial feeding problems but others not. Parents will begin to onotice delay in developmental milestones, as child grows.

1st Symptom: Lack of smiling at 6 weeks and all motor milestones usually delayed. Distinct facial features which although dysmorphic not strikingly. Hypotonia.

3. Diagnosis & 1st Treatment
Blood test only available since 2007 but needs a geneticist to suspect PTHS MRI may find some structural anomalies. Distinct facial features, motor and cognitive delay, hypotonia, lack of speech, breathing regulation anomalies, unstable gait if walking, GI problemsconstipation, reflux myopia, slender fingers, palmar crease, stereotypies

4. Surgery
Some children may need foot surgery if their feet pronate too much. Some may need surgery for undescended testes. Possibly for strabismus

5. Follow-Up
Many children need medication for constipation, reflux, seizures. Some have oxygen for breathing anomalies.

Ideally:
Doctors take parents' concerns seriously early so therapy can begin promptly. Initially this is physiotherapy.

Note: Physiotherapy once doctors agree there is developmental delay. In addition children often receive OT, ST, MT and SIT. Regular follow-up by paediatrician. May have medication for constipation or reflux

Note: Sight and hearing checked. Feet looked at and reviewed regularly for specalist footwear. Back checked for scoliosis. EEG done for base-line. Advice for constipation. OT assessment for equipment

Note:
Hydrotherapy available after surgery and other physiotherapy***!***

Ideally:
Good communication between different specialists and therapists.

Note: SEN schools involvement early on. Respite for family. Help with care in home. Regular access to specialist***!***

Information and European support groups:
www.pitthopkins.org.uk
www.pitthopkins.nl
www.pitthopkins.ch
www.aisph.it

EURORDIS.ORG