Case History

By Dr Keong Mok (Paediatric)

Learning

“ … But we can learn when the knowledge is given to everyone and we can have lots of leaders, and so everyone will be strong not just the ones who have been chosen. Because sometimes, even if you are the leader and you need to be strong, you can get tired.”

Objective – Short ?

History

Question ourself

Examination

Consolidation/Formulate/Management

Case History

Presenting Complaint

5 years 8 months Chinese boy

Concern about:

- Growth – Short
- Motor Skill

Background History

1) Congenital heart disease - clinically stable

- Atrial Septal defect, Ventricular septal defect , Pulmonary valve stenosis, Heart dilatation
- Post repair 2016 (done overseas) - Pulmonary valvoplasty , Repair ASD (Atrial Septal Defect) and dredge of right ventricular outflow tract (RVOT)

2) Previous surgery for bilateral congenital ptosis (eyelid raised)

3) Undescended testes repair – 3 years of age


SHORT ?

Ask yourself from history?

AIM IS TO DETERMINE NORMAL VARIENT vs PATHOLOGICAL …

- Is the child short?
- Child's height velocity (HV)?
- Child's likely (predicted) adult height?
- Pubertal stage?
- Growth pattern on growth chart
- Well or Unwell

Height

- Height was 106.5cm (Z-score -1.64, just above 3rd centile). His upper segment measurements was 57.5cm. Weight today was 19kg (25th centile). No puberty.
- His mother's height was 163 cm and his father's height was 185 cm. Calculated mid- parental height was …
- Height last year (16 months ago) was 103cm - estimated height velocity …
- Both sides of the family (paternal and maternal) are of average height.
- Previous bone age delayed and was advised to observe his growth.

Motor Skill

- Walking around 1 1/2 years of age.
- Slower as compared to other children his age.
- Struggles with the stairs and some of the physical demands at school (exercise).
- Fine motor well – Mature pencil grip. Assistance to do up small buttons.

Additional History…

- Cardiac - Recovery well. Good exercise tolerance and no syncope, recurrence chest infection or shortness of breath.
- Eyes – At 2-3 years old corrective surgery for his eyelids (congenital ptosis). Also has ***amblyopia and he is using*** glasses. Follow up ophthalmology
- Genitals - At 2-3 years old surgery for undescended testes.
- There is no obvious history of headaches or abdominal pain.
- Bowel motions are normal with no obvious blood or mucus.

Family/Social History

- Moved to New Zealand about three years ago.
- No concerns with his learning.
- No medication, no allergies and Immunizations are up to date .
- No family history of developmental delay, genetic disorder or congenital heart disease.

Birth history

- Antenatal scan showed increase “nuchal neck thickness”
- Amniocentesis ?normal
- Born overseas at term . Normal vaginal delivery with a birth weight or 3.2 kilograms .
- No NICU admission.

AIM IS TO DETERMINE NORMAL VARIENT vs PATHOLOGICAL …

Ask yourself from history?

- Is the child short?
- Child's height velocity (HV)?
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- Well or Unwell

Is the child short?
5 years 8 month old chinese boy

- Height was 106.5cm (Z-score -1.64, just above 3rd centile).

The child's height velocity (HV) normal ?

- 4.6cm/ year

Will he reach the child's likely (predicted) adult height?

- 185cm (75th centile)

Puberty?

- Not yet

Well or Unwell?

- Well, but had background cardiac history- went through cardiac surgery

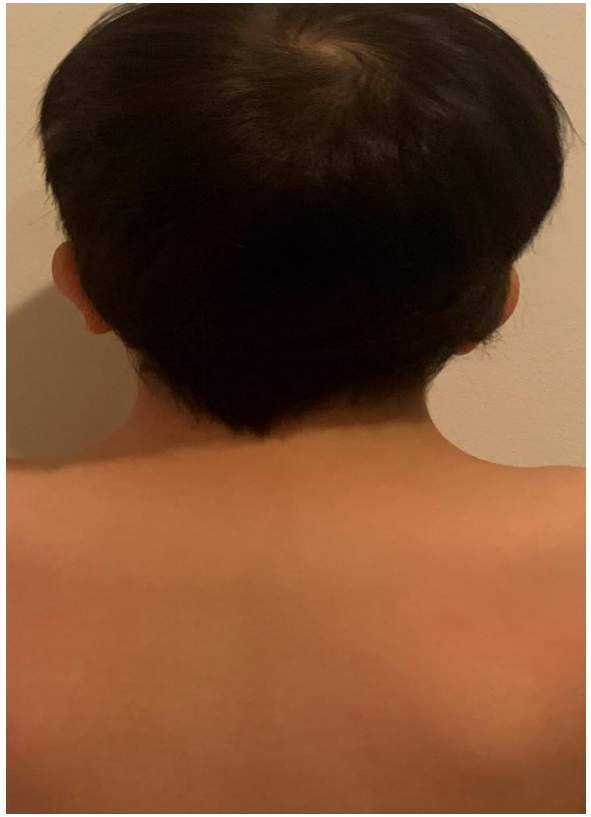
Examination

- Blood pressure 92/56 with a heart rate of 90 and saturation of 96%. Otherwise healthy.
- A mediastinum scar was noted.
- Cardiac/chest normal.
- Abdomen was soft non-tender with no obvious organomegaly.
- No clinodactyly. Normal thyroid, spine, mouth, no obvious rash.
- His neurological examination was normal upper and lower limb neurology. Normal eyes and coordination.

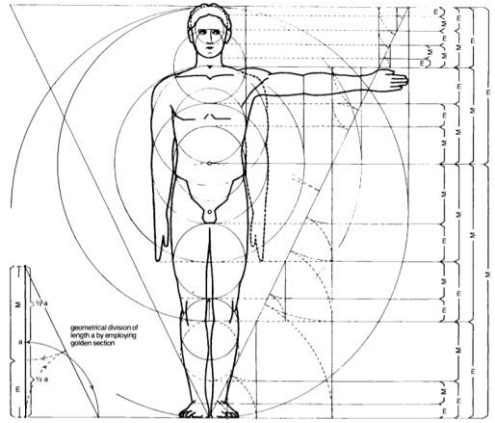
Clinical development

- Normal Language. Normal vision or hearing. Normal fine motor.
- Normal gait and able to run.
- **Heel-toe** walking but was **unable to do this backwards.**
- He could **jump for a short distance**.
- He was only able to **stand on one foot but only for 2 seconds**. **Difficulty hopping on one leg (manage only 2 jumps).**
- He was able to stand up without support from squatting. **Walking down stairs he had to hold on to the rails** and **need** it to put **both feet on one step**.

Clinical picture



- He had some degree of neck webbing and low hairline.
- No clindataly.
- Thyroid examination was normal.
- Limb / Body proportion normal



Consolidate

Height

- Tracking above the 3rd centile (not short stature) but he is below his genetic potential for his height.
- Normal Growth velocity and no puberty
- Investigation

Consolidate

Motor delay

- Some degree of gross motor delay. Normal neurology.
- The rest of his development was normal.

Basic Investigations

- Normal Full blood count
- Normal Liver and kidney function
- Normal Thyroid function tests
- IGF-1 120ng/ml (-0.6 SD)
- Normal Coeliac screen and B12 and folate

- Faecal calprotectin slightly elevated 80 (repeat normal)
- Normal Calcium , phosphate , Vitamin D level
- Normal CK

X-ray wrist (chronological age 5 years 8 month) showed bone age was 5 years old.

Consolidate

Congenital heart disease

- Cardiovascular wise he was clinically well and asymptomatic.
- No follow up here

ECG (23/3/21) : sinus rhythm with partial right bundle branch block with left axis deviation. The R wave in V1 was above the upper limit of normal but there were no other features to suggest hypertrophy. Calculated QTc was 0.36. No previous ECG to compare.

Consolidate

Others

- Eyes : *Amblyopia and he is using* glasses has follow up with ophthalmology.
- Undescended testes repair – stable no follow up

Formulate

Height

- Height just above the 3rd centile (Z-score -1.64)
- Below genetic potential.
- Normal bone age and growth velocity.
- Neck webbing and low hairline

- Background of :

Antenatal increase in nucal fold thickness

Congenital heart disease - clinically stable

Previous surgery for bilateral congenital ptosis (eyelid raised)

Undescended testes repair – 3 years of age

Formulate

Gross motor delay

- Normal neurology
- Other development normal

- Background of :

Antenatal increase in nucal fold thickness

Congenital heart disease - clinically stable

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On the cards

- Potential syndromic cause (?RASopathy) - Noonan Like phenotype.

Management

- Height – Genetics, Endocrine referral
- Motor delay – physiotherapy support
- Cardiac – Cardiology

Diagnosis

- Confirm Noonan's
- Endocrine to consider growth hormone
- Seen by physiotherapy
- Cardiology review – follow up in 3 years time

Wish family all the best

Growth hormone for Noonan's

- **Noonan syndrome**

Subcutaneous injection (35 micrograms/kg daily *or* 1 mg/m^2 daily)

Side effect :

- Allergic reaction, including swelling at the injection site, rash, or hives
- Joint pain (Hip & knee)
- Headaches
- Progression of spine curvature in patients with scoliosis
- Temporary increase in blood sugar levels, which stops when the GH treatment stops
- ? Future increase risk of cancer

References

- https://fpnotebook.com/Endo/Exam/index.htm
- https://dermnetnz.org/topics/noonan-syndrome
- NZF children, (https://nzfchildren.org.nz)