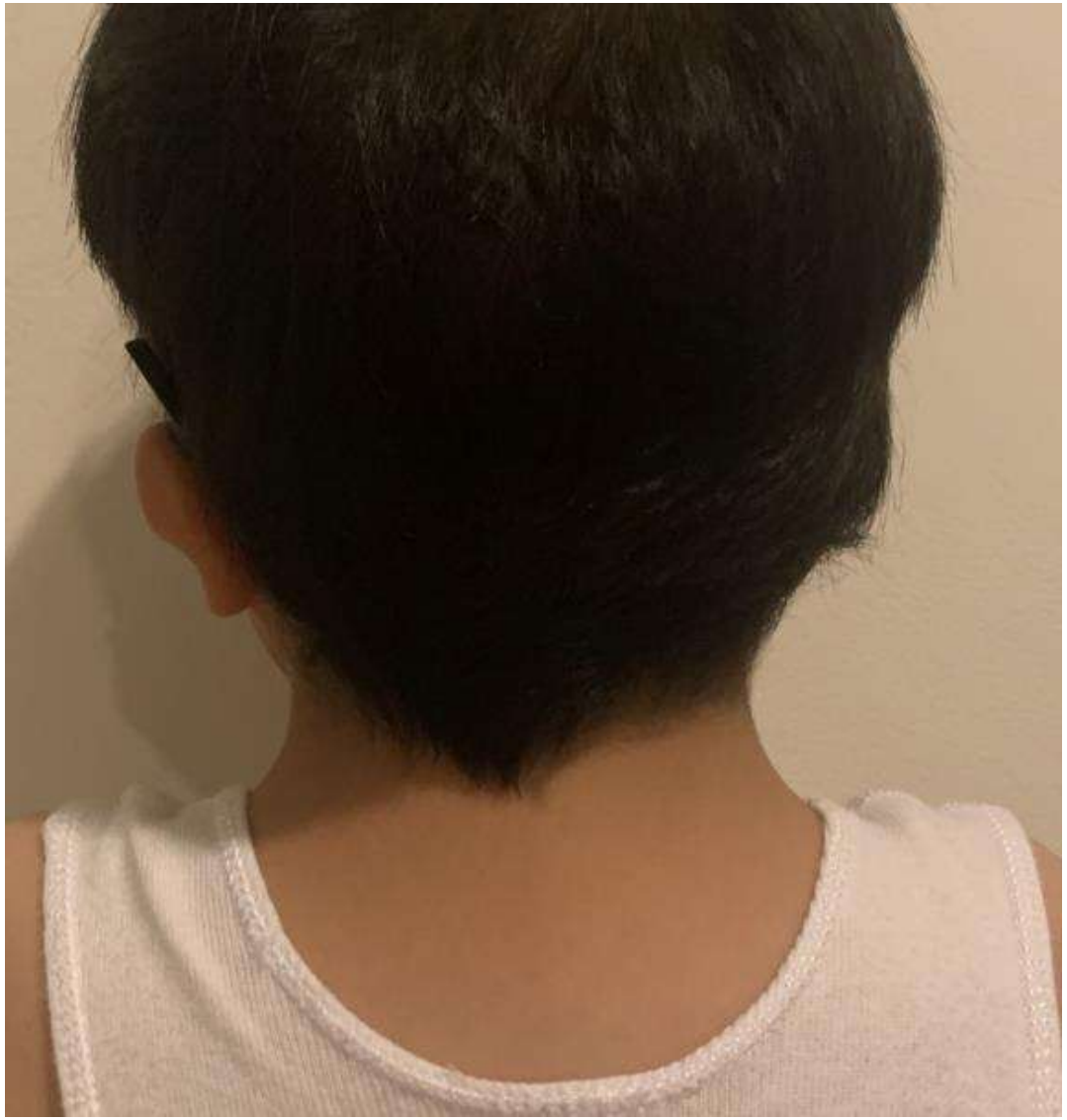

NECK/Head

Examination

- Unable to walk backwards. Jump for a short distance. He was only able to stand on one foot for 2 seconds. He had difficulty hopping on one leg (manage only 1-2 hops)
- Walking downstairs, he had to hold on to the rails and need it to put both feet on one step.
- Normal neurology
- Other development appropriate

QUESTION:

- What are your thought ?

Height/Growth

Motor skill

- What is your plan ?
- Any investigation ?
- Treatment ?

Regarding Master Two's height

Is there a problem? 

Short stature ? 

Puberty? 

Appropriate height velocity ?

Slight below 

Reach genetic potential ? 

Does it matter ?

…

Is there others to suggest pathological Cause ? 

- Child <1st centile
- Abnormally short for family heights
- History/exam suggests chronic illness (especially weight loss more than height loss)
- Abnormal growth velocity (<25th centile)
- Body proportions abnormal
- Dysmorphic features or midline defects

QUESTION:

- What are your thought ?

Height/Growth

Motor skill

- What is your plan ?
- Any investigation ?
- Treatment ?

Investigations

- X-ray wrist (chronological age 5 years 8 month) showed that his bone age was 5 years old.
- Normal Full blood count
- Normal Liver and kidney function
- Normal Thyroid function tests, CK
- IGF-1 120ng/ml (-0.6 SD)
- Normal Coeliac screen and B12 /folate
- Faecal calprotectin repeat normal
- Normal Calcium , phosphate , Vitamin D
- ECG (23/3/21) : Partial right bundle branch block. The R wave in V1 above the upper limit of normal but no other features to suggest hypertrophy.

Diagnosis

- Refer to endocrine, physiotherapy, cardiology and genetics.

Noonan syndrome

Table 6. Causes of pathological growth: Endocrine PICNICS

		Mechanism	Features
Endocrine	Hypothyroidism	Thyroid hormone promotes long bone growth	Weight gain, lethargy, poor school performance, constipation, dry skin, neonatal jaundice, bradycardia
	Growth hormone (GH) deficiency		Cleft lip and palate or midface hypoplasia, neonatal hypoglycaemia, increased fat to lean body mass ratio
	Cushings syndrome	Excess glucocorticoid slows linear growth	Weight gain, cushingoid features
Psychosocial	Deprivation	Decreased GH secretion[7]	
Iatrogenic	Glucocorticoid use		Cushingoid features
	Spinal irradiation	Shortened vertebral bodies	Lower upper segment: lower segment ratio
Chronic illness	Gastrointestinal, eg. inflammatory bowel disease (*Figure 3*) or coeliac disease	Poor nutrition	Loss of weight or failure to gain weight
	Renal, eg. chronic kidney failure or renal tubular acidosis Haematological disorders Complex congenital cardiac disease Cystic fibrosis Metabolic disorders		Features of the underlying disorder
Nutritional		Inadequate nutrition	Loss of weight in absence of other findings Siblings may also be affected
Intrauterine growth retardation	Unknown aetiology or part of a syndrome, eg. Russel-Silver syndrome[8]		Hemihypertrophy, triangular face for Russell-Silver syndrome
Chromosomal	Turner syndrome Down syndrome Prader-Willi syndrome		Features of the syndrome
Skeletal dysplasia	Eg. achondroplasia (autosomal dominant)		Disproportionate short stature Skeletal deformity FH of skeletal dysplasias[9]

https://www.racgp.org.au/getattachment/34d30099-0b3d-4d02-97be-40dd921ad7ea/200509simm.pdf

Conclusion

Conclusion

- Measure, plot and calculate height velocity/mid-parental height
- Understand the principle of height and weight in children
- Examine for Tanner stage
- Looking for features (on examination)
- Bone age and chronic bloods
- ? Genetics
- Refer if unsure

Reference

- GP notebook: https://fpnotebook.com/Endo/
- Growth disorder article: https://www.racgp.org.au/getattachment/34d30099-0b3d-4d02-97be-40dd921ad7ea/200509simm.pdf
- Growth chart : https://apeg.org.au/clinical-resources-links/growth-growth-charts/
- Perkins JM, Subramanian SV, Davey Smith G, Özaltin E. Adult height, nutrition, and population health. Nutr Rev. 2016 Mar;74(3):149-65.
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Thank you