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RARE CAUSE OF SHORT STATURE - CASE REPORT

Paediatrics

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KEYWORDS

BACKGROUND: Isolated growth hormone deficiency is a condition caused by a severe shortage or absence of growthhormone experiencing failure to grow at expected rate and have unusually short stature usually apparent by early childhood. Growth hormone deficiency can either be congenital or acquired Incidence is around 1 in 4000 to 1in 10000. Isolated growth hormone deficiency usually occurs as a result of certain gene mutations. There are three specific gene mutations that can cause isolated GHD. They include: GH1, GHRHR, or BTK genes.

Case report

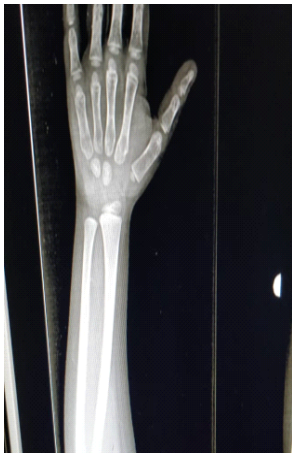
A 11yr old male child 3rd born to 2º consanguineous marriage brought to hospital with chief complaint of not gaining in height according to his age. Child was born by normal vaginal delivery at term with birth weight of 2.7 kg (50th centile).and length 47cm(50th centile) and no nicu admissions. Child have normal increase in height till 3ys of age from then no gain in height. No similar complaints in family. Nutition history and developmental history normal and developemental age is appropriate to chronological age. Child studying 6th class now Intelligence normal.

On general examination Low set ears, depressed nasal bridge, long filtrum present.

Anthrapometry:

	Observed	Expected	Inference
Weight	15 kg	35 kg	
Height	102cm	142cm	

Upper segment/lower segment ratio Normal
Midparent height = 163.5 cm
Target height =164
According to IAP growth chart for boys (5-18yrs) CA > WA> HA> BA
WEIGHT AGE 4 ½ YRS
HEIGHT AGE 4 YRS
BONE AGE 3 YRS
DENTITION DELAYED
SMR STAGE 1. Head circumference normal.
Systemic examination normal.



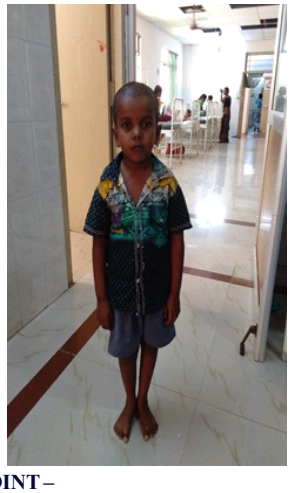


Figure 1.X RAY LEFT WRIST JOINT – ONLY 2 CARPAL BONES PRESENT BONE AGE 3YEARS.

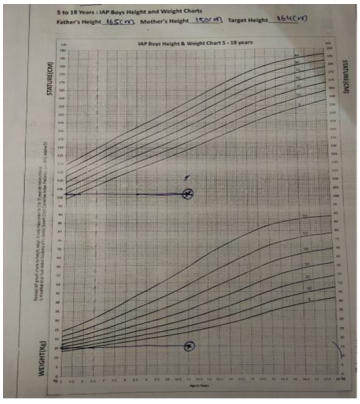


Figure 2. Iap Growth chart 5-18 years for boys. Both weight and height are below 3rd centile.

INVESTIGATIONS:

CBC, Serum calcium, blood glucose normal, immunoglobulin levels normal. Thyroid profile, Vitamin D, ALP, PTH, ABG, serum cortisol – Normal MRI brain normal GH 0.112 ng/ml (low) IGF 1.18 ng/dl (30-300 ng/dl) suggestive of isolated growth hormone deficiency.

Treatment: sub cutaneous Inj.HUMAN Recombinant Growth factor started.

DISCUSSION.

Isolated growth hormone deficiency can be categorized into four groups which are differentiated based on how severe the condition is, the genes involved and the inheritance pattern of the disease Type1a most severe form CAUSED BY absent GH and evident in infancy Type IB produce very low levels of GH characterized by short stature not as severe as in type IA apparent in early to mid-childhood.

Type II have very low levels of GH characterized by short stature not as severe as in type IA apparent in early to mid-childhood.

Type3 similar to type 2 but also have decreased immunity

CONCLUSION:

whenever a child presented with short statutre for long duration in early r mid childhood growth hormone deficiency must to be ruled out. Besides short stature this children have other co morbidities hence timely diagnosis and treatment important.

REFERENCES:

1) Nelson Textbook of Paediatrics- 20th edition.
2) IAP text book of paediatric endocrinology.