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Case Series Of VDDR Type 1A : Underdiagnosed Entity In Children

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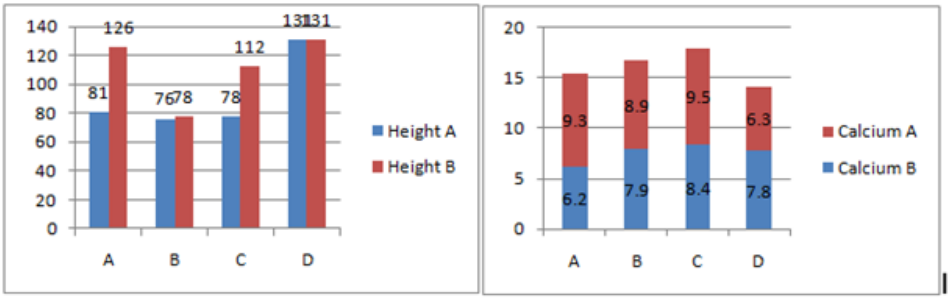
Case Study : Vitamin D-dependent rickets type 1A (VDDR1A) is a rare autosomal recessive disorder caused by deficiency of the CYP27B1 gene which encodes 1alpha hydroxylase which converts 25 hydroxy vitamin D3 to 1,25dihyroxyvitamin D3 which is the active form . VDDR 1B is due to the deficiency of CYP2R1 which encodes 25 hydroxylase which helps in production of 25 hydroxy vitamin D3. Children with VDDR 1A usually become symptomatic in early infancy presenting with typical skeletal signs of rickets, failure to thrive, hypotonia, irritability, seizures or facture as late presentation.VDDR 1B show a milder phenotype and often improves with age.In VDDR 1A 1,25 dihydroxyvitamin D3 level will be low with normal 25 hydroxy vitamin D3 level whereas in type 1B only 25 hydroxy vitaminD3 levels will be low.We are reporting 4 cases of VDDR 1A .The common age of onset was between 1 to 2 years. Out of 4 ,three are male and one female. The commonest clinical symptoms at diagnosis were delayed walking and severe growth retardation .Common examination finding are wrist widening , double malleolus and hypotonia. Investigations revealed all children had hypocalcemia, hypophosphatemia, increased serum alkaline phosphatase and increased parathyroid hormones. All 4 had normal Vitamin D levels as expected in Type 1A supporting our clinical diagnosis. On genetic analysis all children showed CYP27B1 mutations. They were treated with calcitriol, oral calcium supplements and followed up for 2 years. One of four children had poor compliance.We noted biochemical parameters calcium, phosphorus , alakaline phosphatase , parathyroid hormone levels improved parallely with height centile during the course of follow up(Table1).Comparison of Height, Calcium, Vitamin D levels before and after treatment(Figure1).VDDR 1A should be considered in children with rickets who are non responsive to cholecalciferol treatment. Genetic analysis is important for making an accurate diagnosis.

Table 1.png

Table-1 – Comparison of Clinical characteristics and Biochemical profile

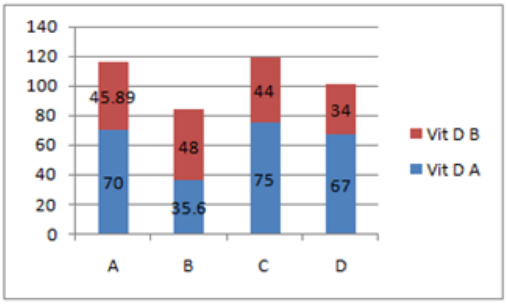
	Case A	Case B	Case C	Case D
Age at presentation	3 years	1 year 4 month	1 year 11 months	2 years
Gender	Female	Male	male	male
Height at presentation	81cm	76cm	78cm	131cm
Physical examination	Delayed dentition Rachitic rosary Harrison sulcus, Double malleolus	Frontal bossing	AF wide open, Frontal bossing, Double malleolus	Scoliosis Right Genu valgum
Calcium (mg/dl)	6.2	7.9	8.4	7.8
Phosphorus(mg/dl)	3.8	1.92	2.4	3.7
ALP (IU/ml)	1611	2000	2965	338
PTH (pg/ml)	632	461	472	366
Vitamin D (ng/ml)	70	35.56	>70	67
Post treatment				
Height	126	78	112	131cm
Calcium(mg/dl)	9.3	8.9	9.5	6.3
Phosphorus(mg/dl)	4.7	2.81	4.64	4.43
ALP(IU/ml)	499	1358	312.8	528.6
PTH(pg/ml)	105	371.4	53.5	252.8
Vitamin D (ng/ml)	45.89	48	44	34.06

Table 1.png



Comparison of Height before and after treatment

Comparison of Calcium levels before and after treatment



Comparison of Vitamin D levels before and after treatment

A- After treatment

B- Before treatment

Figure-1 Comparison of Height, Calcium, Vitamin D levels before and after treatment