

Results

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The study was conducted on 45 patients, all of whom exceeding the age of 2 years and each was showing clinical and laboratory evidences of rickets. It included as well 15 non rachitic children (as proved by clinical and laboratory evidences) that have been used as the control group.

Our 45 patients exhibiting late rickets included 18 patients (40% of total number of patients) having vitamin D deficiency rickets, 8 patients having renal osteodystrophy (18%), 4 patients having vitamin D dependent rickets (type I) (9 %), 6 patients having familial hypophosphatemic rickets (13 %), 8 patients having Fanconi syndrome (18%), and a single case having end organ resistance to active vitamin D.

As regards sex, our group of patients having vitamin D deficiency rickets (18 patients) included 9 males and 9 females (50% for each), while that of renal osteodystrophy (8 patients) included 5 females (62.5%) and 3 males (37.5%), that of vitamin D dependent rickets (type I) (4 patients) included 4 males (100%), that of familial hypophosphatemic rickets (6 patients) included 5 males ($83\frac{1}{3}$ %) and one female ($16\frac{2}{3}$ %), that of Fanconi syndrome (8 patients) included 5 males (62.5 %) and 3 females (37.5 %), while the single case of end organ resistance to the study was a female patient.

As regards age, cases having vitamin D deficiency rickets were ranging between 2 and 4 years with a mean of 2.8

years for that group, while cases having renal osteodystrophy were ranging in age between 3.2 and 13 years with a mean of 5.2 years, cases with vitamin D dependent rickets (type I) were ranging in age between 2.2 and 4 years with a mean of 3.2 years, cases with familial hypophosphatemic rickets were ranging between 2.4 and 5 years with a mean of 3.3 years, cases with Fanconi syndrome were ranging between 2.9 and 7 years with a mean of 3.9 years, while the single case of end organ resistance to active vitamin D was 8 years old by the time of examination.

Statistical analysis of the former results showed that the differences between the mean of ages (on examination) of the vitamin D deficiency group and each of that of renal osteodystrophy and end organ resistance to active vitamin D were very highly significant, while the difference between the mean of vitamin D deficiency rickets and that of Fanconi syndrome was significant. The differences between vitamin D deficiency rickets and other groups; namely: vitamin D dependent rickets (type I) and familial hypophosphatemic rickets as regards age by the time of examination were statistically non significant.

The group of patients having vitamin D deficiency rickets has had an onset of rachitic manifestations that ranged between $\frac{1}{2}$ -1 $\frac{1}{2}$ years with a mean of 0.97 years, while those patients having renal osteodystrophy have had their onset of rachitic manifestations by the age of 1 $\frac{1}{2}$ to 10 years,

with a mean of 3.6 years, those having vitamin D dependent rickets (type I) were exhibiting a range of $\frac{1}{2}$ to $1\frac{1}{2}$ years with a mean of 1 year, those having familial hypophosphatemic rickets were ranging between 1 and 2 years with a mean of 1.5 years, those patients with Fanconi syndrome were ranging between 1 and 2 years with a mean of 1.2 years, while the single case of end organ resistance to active vitamin D has had clinical manifestations that dated back since the age of one year.

Statistical analysis of these results showed a highly significant difference between the means of age of onset in vitamin D deficiency group and renal osteodystrophy group, while there were no statistical significance between the age of onset of vitamin D deficiency rickets and the four other groups.

Throughout our study 9 children out of our 45 cases exhibiting late rickets were exceeding the age of 4 years (20%) including 5 cases with renal osteodystrophy (out of 8 patients presenting in the study) i.e (62.5 %), 2 cases of Fanconi syndrome, and one case accounting for each of familial hypophosphatemic rickets and end organ resistance to vitamin D.

Regarding the weight of patients included in our study, none of them was exceeding the 95th centile for age. It is even note worthy that none of our patients was exceeding the 50th centile for age. An under weight was a finding commonly met with throughout the study, where a remarkable number of

patients (68.9 %) were found to have their weights below the 5th centile for age. The prevalence of under weight in our patients having late rickets was found to be 61.1 % in the vitamin D deficiency group and 50% in the renal osteodystrophy group, 75% in vitamin D dependant rickets, 67 % in familial hypophosphatemic rickets, 75 % in Fanconi syndrome group, while the single case of end organ resistance to vitamin D was having her weight below the 5th centile for age by the time of examination. There were no statistically significant differences between the various study groups as regards the prevalence of under weight in them.

As regards height, it was noticed that 13 of our 18 patients having vitamin D deficiency rickets (72.2 % of them) were having their height below the 5th centile for age. Such an under stature was found in all cases having rickets due to causes other than vitamin D deficiency (100 % of cases having renal osteodystrophy, vitamin D dependent rickets (type I), familial hypophosphatemic rickets, Fanconi syndrome, and end organ resistance to active vitamin D).

As regards consanguinity, history of positive consanguinity was obtained in 4 cases out of our 18 patients having vitamin D deficiency rickets (22.2 %). Such consanguinity was positive in one case out of 8 patients with renal osteodystrophy (12.5 %), in all cases of vitamin D dependent rickets (100 %), in 4 out of the 6 cases of familial hypophosphatemic rickets (66.7 %), 4 out of the 8 patients of Fanconi syndrome

(50 %), and in the case of end organ resistance to active vitamin D.

Family members showing similar rackitic manifestations could be detected in 2 out of our 18 cases showing vitamin D deficiency rickets, in one case out of the 8 cases of renal osteodystrophy, non of cases of vitamin D dependent rickets (type I), in one case out of the 6 cases of familial hypophosphatemic rickets, and in one case out of the 8 cases of Fanconi syndrome.

Although rachitic manifestations could be detected in all of our patients, evaluation of our findings pointed to some clinical aspects where variable expressivity of rachitic manifestations was encountered.

Frontal bossing was evident in 8 patients out of the 18 cases having vitamin D deficiency rickets (44.4 %), in 3 patients out of our 4 patients having vitamin D dependent rickets (75%), in 3 cases out of our 8 cases with Fonconi syndrome (37.5 %), and in the single case having end organ resistance to active vitamin D. It could not be detected in any of our cases having renal osteodystrophy or those having familial hypophosphatemic rickets.

Harrison sulcus could be detected in 6 cases out of the 18 patients with vitamin D deficiency rickets (33.3 %), in a single case out of our 8 cases having renal osteodystrophy (12.5 %), in 2 cases out of 8 having Fanconi syndrome (25 %), and in the single case of end organ resistance to active

vitamin D, while it could not be detected in any of our patients having vitamin D dependent rickets (type I) or in familial hypophosphatemic rickets.

Rachitic rosary could be detected in 6 cases out of our 18 cases of vitamin D deficiency rickets (33.3 %), in 3 cases out of 8 with renal osteodystrophy (37.5 %), in one case out of the 4 cases having vitamin D dependent rickets (25 %), in 5 cases out of the 8 cases of Fanconi syndrome (62.5 %), and in the single case of end organ resistance to active vitamin D. Rachitic rosary could not be detected in any of the cases of familial hypophosphatemic rickets.

Rachitic chest manifestations that have been encountered throughout our study in their order of frequency were: rachitic rosary in 16 cases (35.5 % of the total group of patients), Harrison sulcus in 10 cases (22.2 % of the total), while pigeon chest deformity could be detected only in a single case belonging to the vitamin D deficiency type and another case of marked chest deformity was found among cases having Fanconi syndrome.

- Marfan sign could be detected in each of our 45 cases except for a single case of renal osteodystrophy in whom the condition started by the age of 10 years.

- Either genu varum or genu valgum could be detected in each of our 45 cases exhibiting rachitic manifestations except for one case belonging to the renal osteodystrophy group.

- Genu varum was much more commonly detected than genu valgum in all groups except for the group of renal osteodystrophy in which genu valgum could be detected in 6 out of its 8 cases (75 %).

- Delayed walking (after the age of 2 years) as one of the rachitic manifestations was met with in 17 out of the 45 rachitic cases (37.8 %). Delayed walking was encountered in all types of rickets with an incidence of 75% in vitamin D dependent rickets, 37.5 % in Fanconi syndrome, 33.3 % in either of vitamin D deficiency rickets and familial hypophosphatemic rickets, 25 % in renal osteodystrophy, and in that single case of end organ resistance to active vitamin D. Except for 4 cases, none of our studied cases was unable to walk after the age of 3½ years. These 4 cases included a single case of vitamin D deficiency rickets (who was 4 years old), a single case of vitamin D dependent rickets (who was 4 years old), a single case of Fanconi syndrome (who was 7 years old), and the single case of end organ resistance to active vitamin D (who was 8 years old).

- Hypotonia which could be concluded from pot belly abdomen with or without umbilical hernias and hypotonia in extremities as well as delayed milestones of motor development (including primary inability to walk), correctable kyphosis, and others was found in 28 cases out of our 45 studied cases (62.2 %). Hypotonia was found in 13 out of 18 cases of vitamin D deficiency rickets (72.2 %), in 3 out of 8 cases with renal

osteodystrophy (37.5 %), in 3 out of 4 cases with vitamin D dependent rickets (75%), in 6 out of 8 cases with Fanconi syndrome (75%), in 2 out of 8 cases with familial hypophosphatemic rickets (25%), and in the single case of end organ resistance to active vitamin D during examination.

- Anterior fontanelle was widely open in 2 cases in our study. The first belonging to the vitamin D dependent rickets whose age was $3\frac{1}{2}$ years, and the other case had Fanconi syndrome whose age was 3 years. It was not found in any of the remaining cases including all those of late vitamin D deficiency rickets.

- Pathological fractures were encountered only in 2 cases belonging to the group of Fanconi syndrome. One had fracture femur, and the other had fracture femur plus fracture of the humerus bone. Such fractures were not encountered in other groups.

Serum alkaline phosphatase activity:

Regarding serum alkaline phosphatase activity-which had to be higher than normal in each of our patients being used as a diagnostic laboratory measure for the presence of rickets-its level was found to range between 8 and 17 King-Armstrong units with an average of 12.53 K.A.U. in the control group, while it was found to have a range between 16 and 75 K.A.U. with a mean of 32.06 K.A.U. in vitamin D deficiency rickets group, a range between 24 and 75 K.A.U. with a mean of 46 K.A.U. in the group of renal osteodystrophy, a range between 42 and

60 K.A.U. with a mean of 51.75 K.A.U. in cases of vitamin D dependent rickets, a range between 30 and 90 K.A.U. with a mean of 46 K.A.U. in cases of familial hypophosphatemic rickets, a range between 30 and 96 K.A.U. with a mean of 51.63 K.A.U. in cases of Fanconi syndrome, while the single case of end organ resistance to active vitamin D was having a level of 45 K.A.U. It is note worthy to mention that statistically significant differences would be found between all rachitic groups at one hand and the control group at the other hand as regards alkaline phosphatase activity. It is also important to mention that the mean of alkaline phosphatase activity was found to be highest in the vitamin D dependent rickets group, next high in Fanconi syndrome, next in both renal osteodystrophy and familial hypophosphatemic rickets, next in the single case of end organ resistance to active vitamin D, and was found to be lowest in the vitamin D deficiency rickets group with no statistically significant differences among means in different rachitic groups.

- Regarding serum calcium level, it was found to lie within the standard normal range for serum calcium (8.5-10.5 mg/d.L.) in each of the control group cases with a range of 8.75-10 mg/d.l. with a mean of 9.5 mg/d.l.

The average serum calcium level was found to be lower than that of the control group in each of different examined rachitic groups except for that of Fanconi syndrome, a difference which was found to be of statistical significance.

Vitamin D deficiency cases exhibited a normal serum calcium level in 9 cases (50%) and a level lower than normal in the remaining 9 cases (50%). The latter 9 cases included a single case of a serum calcium level of 8.13 mg/d.L. (Slightly lower than normal) and other 8 cases in whom serum calcium level ranged from 6.25 to 7.5 mg/d.L. None of the vitamin D deficiency rickets cases had a serum calcium level lower than 6.25 mg/d.L. The mean serum calcium level for all cases of vitamin D deficiency rickets (which was found to be 8.3 mg/d.L.) was found to be statistically significantly lower than of the control group and that of the Fanconi syndrome group. It was also found to be statistically significantly higher than its corresponding mean of the vitamin D dependent rickets group, while no statistically significant difference was found between the average serum calcium level for vitamin D deficiency group and those corresponding means for renal osteodystrophy, familial hypophosphatemic rickets, and that single case of end organ resistance to active vitamin D.

Regarding the group of renal osteodystrophy serum calcium level was found to range between 5 and 8.75 mg/dL with a mean of 7.5 mg/dL; that was found to be lower than its corresponding average serum calcium for the vitamin D deficiency group (without statistically significant difference). Regarding the group of familial hypophosphatemic rickets serum calcium level was ranging between 7.5 mg/dL and 9.38 mg/dL with a mean of 8.69 mg/dL which is slightly higher than corresponding mean of vitamin D deficiency rickets group

with no statistically significant difference.

Serum calcium level was found to be 5 mg/dL in each of the cases of vitamin D dependent rickets, a level which was found to be statistically significantly lower than any other studied group.

Regarding Fanconi syndrome cases, serum calcium level was ranging between 8.13 and 12.5 mg/dL with a mean of 9.61 mg/

dL a value which is statistically significantly higher than its corresponding mean of any of the other rachitic groups and is higher than its corresponding mean of the control group with no statistically significant difference between them.

As regards serum phosphorus, each of our studied rachitic groups except for that of renal osteodystrophy have had an incidence of 100 % of serum phosphorus level, that is lower than the standard range (4.5-6.5 mg/dL) and than that of the control group (4.25-5.75 mg/dL). Such a difference was found to be of statistical significance.

Serum phosphorus levels were found to be statistically significantly lower in patients belonging to the familial hypophosphatemic rickets and Fanconi syndrome groups in whom serum phosphorus levels were found to range between 2.1 and 3.25 mg/dL with a mean of 2.68 mg/dL and between 2.5 and 3.6 mg/dL with a mean of 2.99 mg/dL respectively than that of other groups in whom serum phosphorus level was ranging

between 2.75 and 4.2 mg/dL with a mean of 3.55 mg/dL in the group of vitamin D deficiency rickets, between 2.5 and 4 mg/dL. with a mean of 3.44 mg/dL in vitamin D dependent rickets group, and was found to be 3.4 mg/dL in the single case of end organ resistance to active vitamin D.

A statistically significantly higher serum phosphorus level was found in those patients belonging to renal osteo-

dystrophy (the mean of which was 7.62 mg/dL) than those patients belonging to other rachitic groups as well as the control group. Although it is evident that 50 % of cases belonging to the renal osteodystrophy group were having a high normal serum phosphorus level when the standard serum phosphorus level is considered for such evaluation, however it is note worthy to mention that all cases belonging to that group have had a serum phosphorus level that was found to be higher than the high limit obtained from our control group.

Regarding urinary calcium, control cases were found to have an average of 53.87 mg/24 hours urine, vitamin D deficiency cases had an average of 46.71 mg/24 hours urine, those having renal osteodystrophy had a mean of 37 mg/24 hours, vitamin D dependent rickets group had a mean of 80.69 mg/24 hours, familical hypophosphatemic rickets group had an average of 42 mg/24 hours urine, Fanconi syndrome group had a mean

resistance to active vitamin D had a level of 50 mg/24 hours urine.

Comparing the means of various studied groups, all of them were found to lie within the normal standard range for urinary calcium which was stated to be less than 300 mg/24 hours (Tietz and Finley, 1983). However, it is note worthy that the obtained means for urinary calcium in Fanconi syndrome

and as well in cases of vitamin D dependent rickets were found to be higher than the mean of our control group and the differences were statistically significant. There were no statistically significant differences between other means of the different groups of rickets and that of our control group.

Regarding urinary phosphorus the obtained means were 100 mg/24 hours urine in the control group, 187.52 mg/24 hours urine in vitamin D deficiency rickets group, 174.88 mg/24 hours urine in renal osteodystrophy group, 179.69 mg/24 hours urine in vitamin D dependent rickets group, 795.33 mg/24 hours urine in familial hypophosphatemic rickets group, 776.75 mg/24 hours urine in Fanconi syndrome, and 267 mg/24 hours urine in the single case of end organ resistance to active vitamin D. In spite of the fact that all the means of rachitic groups are statistically significantly higher than the mean of the control group, only the means of familial hypophosphatemic rickets group and Fanconi syndrome group are lying

was stated to be 10-20 mg/kg body weight/24 hours (For far and Arneil, 1984). These two values were found to be statistically significantly higher than all the means of other rachitic groups as well as the control group.

Urinary glucose mean values were found to lie within the normal standard range in the control group as well as all of rachitic groups except for Fanconi syndrome group being 47.73 mg/24 hours in the control group, 69.59 mg/day in vitamin D deficiency rickets, 44.33 mg/day in renal osteodystrophy, 56.45 mg/day in vitamin D dependent rickets, 57.13 mg/day in familial hypophosphatemic rickets, and 68mg/day in the single case of end organ resistance to active vitamin D. In Fanconi syndrome, the mean of urinary glucose was found to be 1326.25 mg/day; a value which is statistically significantly higher than that of the control group as well as each of other rachitic groups.

Regarding urinary proteins, mean values of the control group, vitamin D deficiency group, vitamin D dependent rickets group, familial hypophosphatemic rickets group, as well as in the single case of end organ resistance to active vitamin D were found to be within the normal standard value being 0.13, 0.12, 0.15, 0.14, and 0.15 gram/24 hours urine respectively. In renal osteodystrophy and Fanconi syndrome groups the means of urinary protein were found to be higher than the control group as well as the standard normal average being 0.21 gram/day in renal osteodystrophy and 0.23 gram/day in Fanconi

syndrome. These 2 values were found to be statistically significantly higher than the means of the control group and other rachitic groups.

Aminoacid chromatography in urine was found to be normal in each of our control cases and in 34 out of our 45 rachitic patients. Generalized aminoaciduria was found in 2 cases of the vitamin D deficiency rickets, in one case of renal osteodystrophy, and in the 8 cases of Fanconi syndrome, 3 of which were showing generalized aminoaciduria with excess cystine.

Regarding creatinine clearance, the mean of the control group was 95.33 ml/minute/1.73 m² surface area, that of the vitamin D deficiency group was 90.32 ml/minute/1.73 m² surface area, that of vitamin D dependent rickets was 87.17 ml/minute/1.73 m² surface area, that of familial hypophosphatemic rickets was 86.55 ml./minute/1.73 m² surface area, and that of the single case of end organ resistance to active vitamin D was 82ml./minute/1.73m² surface area. All of these values are found to be within normal standard values. The mean value of creatinine clearance in the group of renal osteodystrophy was 15.7 ml./minute/1.73m² surface area, that of Fanconi syndrome was 61.66 ml./minute/1.73m² surface area. These 2 values were found to be statistically significantly lower than that of the control group.

Urine aminoacids chromatogram for seven cases six of whom belonging to Fanconi syndrome and showing generalized aminoaciduria and the seventh is belonging to the control group showing normal urinary aminoacids.

Clinical and laboratory data of the cases of control group (A)

Case no.	Sex	Age (yr)	Wt. (Kg.)	Wt. on Centile curve	Ht. on Centile curve	Serum Ca		Serum P	Alk. Phosphatase	Creat. Cl		Urine Ca		Urine P		Urine glucose	Urine ptn														
						mg%	m.mol/L.	mg%	L.	K.A.U.	I.U./L.	ml/min/1.73m ²	mg/d.	m.mol/d.	mg/d.	m.mol/d.	mg/d.	m.mol/d.	gram/day												
1	♂	7	19	10th	113	5th	8.75	2.19	5	1.61	10	71	105	55	1.38	96	3-1	35	0.19	0.1	Normal										
2	♂	5	17	25th	105	25th	8.75	2.19	4.75	1.53	13	92.3	95	47	1.18	82	2-65	32	0.17	0.2	Normal										
3	♂	9	25	25th	125	10th	10	2.5	4.75	1.53	16	113.6	89	63	1.58	70	2-26	30	0.17	0.1	Normal										
4	♂	3	14	50th	94	50th	10	2.5	5.25	1.69	14	99.4	110	38	0.95	55	1-78	64	0.36	0.1	Normal										
5	♀	3 1/2	13	10th	96	25th	9.38	2.34	5	1.61	11	78.1	117	30	0.75	120	3-87	64	0.36	0.15	Normal										
6	♂	8	25	50th	130	75th	10	2.5	5.25	1.69	12	85.2	92	56	1.41	78	2-51	42	0.23	0.15	Normal										
7	♀	2	11	50th	84	25th	9.38	2.34	4.75	1.53	11	78.1	103	59	1.48	180	5-81	27	0.17	0.1	Normal										
8	♀	2	10	5th	82	10th	10	2.5	4.25	1.37	13	92.3	98	39	0.98	140	4-52	57	0.32	0.15	Normal										
9	♂	7	18	5th	113	5th	8.75	2.187	4.37	1.41	11	78.1	87	84	2.11	165	5-33	73	0.41	0.2	Normal										
10	♀	9	24	25th	123	10th	10	2.5	5.75	1.86	15	106.5	94	60	1.5	74	2-39	88	0.49	0.1	Normal										
11	♂	2 1/2	13	50th	86	10th	9.38	2.34	4.75	1.53	17	120.7	83	72	1.8	64	2-07	35	0.19	0.15	Normal										
12	♂	3 1/2	14	25th	93	5th	10	2.5	4.25	1.37	12	85.2	78	68	1.7	78	2-51	63	0.35	0.1	Normal										
13	♀	5	17	50th	105	25th	8.75	2.19	5	1.61	16	113.6	92	32	0.8	70	2-26	47	0.26	0.1	Normal										
14	♀	4	15	50th	101	50th	10	2.5	4.75	1.53	8	56.8	86	48	1.2	166	5-36	29	0.16	0.2	Normal										
15	♀	3 1/2	14	25th	95	25th	9.38	2.34	5.25	1.69	9	63.9	102	57	1.43	62	2-0	64	0.36	0.15	Normal										
Mean 4.9																		9.5	2.38	4.87	1.57	12.53	88.99	95.53	53.87	1.35	100	3.23	47.73	0.26	0.13
St. devi.																		0.54	0.14	0.41	0.13	2.67	18.96	10.44	15.1	0.38	42.7	1.38	19.1	0.11	0.04
St. error																		0.14	0.04	0.11	0.03	0.69	4.9	2.7	3.9	0.1	11.03	0.36	4.93	0.03	0.01

Table no. 1.

Clinical data of Cases of Vitamin D deficiency rickets(B₁)

Clinical Manifestations of rickets or related conditions														
Height (cm)	Age (yr)	Previous Vit. D intake	Family history (Years)	Boiling of Frontal bone	Delayed or defective Teeth	Harrison's Sulcus	Chest rosary beads	Enlarged wrist	(+)ve X-ray	Genia	Valgum	Delayed walking	Hypotonia	Others
76	<5th	1	irrele.	-	+		+	+	+			+	+	
87	<5th	1	(+)ve consang.	-		+		+	+			+	+	
77	<5th	1	Recent 2 inj.	irrele.				+	+			+	+	
81	<5th	1½	" 2 inj.	"				+	+				+	
81	<5th	1	" 2 inj.	"	+		+	+	+					
82	10th	½	Recent consang. 2 inj.	(+)ve consang.				+	+				+	
75	<5th	¾-1	(+)ve consang.	-		+		+	+				+	Pigeon Chest deformity
80	<5th	1½	rachitic sister 2 inj.	rachitic sister		+		+	+			+	+	Kyphosis.
71	<5th	¾-1	irrele.	-				+	+			+	+	Coming from Sudan (Black).
82	5th	1½	" 3 inj.	"				+	+			+	+	Kyphosis.
77	<5th	1½	has a twin	irrele.	+	+		+	+			+	+	
81	<5th	¾-1	" 1 inj.	"				+	+			+	+	
71	<5th	¾-1	" 2 inj.	"		+		+	+			+	+	
94	10th	1½	"	"				+	+			+	+	
87	10th	1	"	"				+	+			+	+	
84	5th	1	rachitic brother 1 inj.	rachitic brother				+	+			+	+	
76	<5th	1-½	(+)ve consang. 1 inj.	(+)ve consang.				+	+			+	+	
69	<5th	1	irrele. Recent 2 inj.	irrele.	4 22%	6 33%	6 33%	16 100%	18 100%	1 100%	0 0%	6 33%	13 72.2%	

6.8%

27.7% 0.97

Normal

27.7% C.97 Normal

6.6% normal

Clinical data of Cases of renal osteodystrophy										Clinical manifestations of rickets or related conditions									
Height or length (cm.)	Weight or surface area (kg. or sq. m.)	Age or sex	Family history	Previous Vit. D intake	Bossing of frontal bone	Delayed or defective Teeth	Harrison sulcus	Chest rosary beads	Enlarged wrist	(+)ve Marras Sten	Genium Sten	Genium Sten	Delayed walking	Hypotonia	Others				
84	< 5th	1%	irrelev.	3 inj.	+			+	+	+			+	+	Obstructive uropathy				
80	< 5th	2%	irrelev.	3 inj.			+	+	+	+			+	+					
80	< 5th	3%	(+)ve consang. tions	3 injections											Carpopedal spasms				
119	< 5th	10	a foundling	-					+										
78	< 5th	3	irrelev.	-					+	+									
87	< 5th	2%	rachitic causins	-					+	+					Recurrent Nephrotic Syndrome.				
87	< 5th	2%	irrelev.	3 inj.				+	+	+				+					
86	< 5th	3%	-	-					+	+									
%	0%	3.6			0	1	1	3	6	7	1	1	2	3					
	Normal				12.5%	12.5%	12.5%	37.5%	75%	87.5%	12.5%	12.5%	75%	37.5%					

Clinical data of Cases of 19X familial hypophosphatemic rickets (B₄)

Clinical manifestations of rickets or related conditions										Others									
Sex	Age (Years)	Wt. (Kg.)	Wt. on centile curve	Height or Length (Cm.)	Ht. or Length on centile curve	Age or Onset (Years)	Family History	Previous Vit. D intake	Bossing of frontal bone	Delayed or defective teeth	Harrison's sulcus	Chest rosenberg beads	Enlarged wrist	(+)ve X-ray	Marfan sign	Genu Varum	Genu Valgum	Delayed walking	Hypotonia
♂	5 1/2	9	< 5th	82	< 5th	1	(+)ve consang.	3 inj.											
♂	7 1/2	11	< 5th	78	< 5th	1 1/2	(+)ve consang.	3 inj.											
♂	6 3/4	14	50th	84	< 5th	2	(-)ve	3 inj.											
♂	4 1/2	10	< 5th	75	< 5th	1	rachitic system	3 inj.											
♂	3 1/2	13	25th	81	< 5th	1 1/2	(+)ve consang.	3 inj.											
♂	5	14	< 5th	82	< 5th	2	(+)ve consang.	6 inj.											
	3.3	33%	33%		0%	1.5			0	0	0	0	6	6	6	4	2	2	2
									0%	0%	0%	0%	100%	100%	100%	100%	66.6%	33%	33%
			Normal																

Table no. 6.

Clinical data of the case of rickets due to

Table no. 7.

of vitamin B deficiency rickets (B₁)

Table no. 8.

Table no. 8.

Laboratory data of cases of renal osteodystrophy (B₂)

Laboratory data of cases of renal osteodystrophy 1972															
Serum Ca	Serum P	Alkaline phos.	Creat.Cl.	Urine Ca	Urine P	Urine Glucose	Urinary Ptn	Aminoacid chromatogram:	Others						
mg% m.mol/L.	mg% m.mol/L.	K.A.U. I.U./L.	ml./min/1.73m ²	mg/day m.mol/day	mg/day m.mol/day	mg/day m.mol/day	mg/day	g/day							
1.5	1.88	6.25	2.02	75	532.5	15.25	46.5	1.16	112.5	3.63	27	0.15	0.2	Normal	Obstructive Uropathy
1.5	1.88	6.5	2.1	75	532.5	28.1	45	1.13	116	3.75	74.2	0.41	0.15	Normal	
3.75	2.19	5.7	1.84	40	284	15.3	40.5	1.01	135	4.36	43.2	0.25	0.2	Normal	
7.5	1.88	14	4.52	28	198.8	2.4	51.75	1.29	286	9.23	23	0.13	0.4	Normal	
8.75	2.19	6.7	2.16	42	298.2	7.43	36	0.9	160	5.17	19.2	0.11	0.15	Normal	
8.75	2.19	6.75	2.18	42	298.2	29.1	44	1.1	300	9.59	52.8	0.29	0.15	Normal	genamino-aciduria
5	1.25	8.75	2.83	24	170.4	10.01	13.5	0.34	127	4.1	70.2	0.39	4(±)	Normal	Nephrotic Synd.
6.25	1.56	6.3	2.03	42	298.2	13.76	18.75	0.47	162.5	5.25	45	0.25	0.2	Normal	
7.5	1.88	7.62	2.46	46	326.6	15.17	37	0.93	174.88	5.65	44.33	0.25	0.21		
1.34	0.33	2.73	0.88	19.18	136.15	9.36	13.73	0.34	75.23	2.43	20.78	0.12	0.09		
0.47	0.12	0.96	0.31	6.78	48.08	3.31	4.85	0.12	26.6	0.86	7.35	0.04	0.03		

this result was excluded in statistical analysis.

Table no. 9.

Laboratory data of cases of ly hypophosphatemic rickets (B₄)

Case no.	Serum Ca		Serum P		Alkaline Ph.		Cr. clearance		Urine Ca		Urine P		Urine Glucose		Urinary Ptn		Aminoacids
	mg%	m.mol/L.	mg%	m.mol/L.	K.A.U.	I.U./L.	ml/min/1.73m ²	mg/day	m.mol/d.	mg/d.	m.mol/d.	mg/day	m.mol./d.	gram/d.	Ptn		
46	7.5	1.88	2.75	0.89	30	213	99.6	40.5	1.01	1040	33.58	48	0.27	0.15			Normal
47	8.75	2.19	2.25	0.73	36	255.6	138.3	27	0.68	747	24.12	39.6	0.22	0.15			Normal
48	9.38	2.34	3	0.97	33	234.3	61.5	48	1.2	545	17.6	32.4	0.18	0.1			Normal
49	8.75	2.19	3.25	1.05	60	426	73.1	36	0.9	874	28.22	79.2	0.44	0.1			Normal
50	8.75	2.19	2.75	0.89	27	191.7	59.9	37.5	0.94	599	19.34	75.6	0.42	0.2			Normal
51	9	2.125	2.1	0.68	90	639	87.4	63	1.58	967	31.22	68	0.38	0.15			Normal
Mean	8.69	2.17	2.68	0.87	46	326.6	86.55	42	1.05	795.33	25.68	57.13	0.32	0.14			
St. devia	0.36	0.09	0.44	0.14	24.57	174.43	28.93	12.33	0.31	199.53	6.44	19.74	0.11	0.038			
St. error	0.15	0.04	0.18	0.06	10.03	71.21	11.81	5.03	0.13	81.46	2.63	8.06	0.04	0.015			

Table no. 11.

Laboratory data of cases of Fanconi syndrome (B5)

Laboratory												Urinary		Aminoacids Chromatogram	
Serum Ca		Serum P		Alk. Phosph.		Cr. clear- ance		Urine Ca		Urine P		Urine Glucose			Ptn
mg L.	mmol/ L.	mg% m.mol/ L.	mg% m.mol/ L.	K.A.U.	I.U./L	ml./min/ 1.73m ²	mg/day	mmol/ day	mg/day	mmol/ day	mg/day	mmol/ day	mmol/ day		gram/day
10	2.5	2.5	0.81	67	475.7	104.6	94.5	2.36	778	25.12	1040	5.77	0.15	0.15	Gen. aminoaciduria with excess cystine
8.75	2.19	2.75	0.89	30	213	35.2	48	1.2	921	29.74	765	4.25	0.15	0.15	Gen. aminoaciduria.
11.25	2.81	3.25	1.05	30	213	49.1	61.88	1.55	650	20.99	621	3.45	0.3	0.3	Gen. aminoaciduria with excess cystine
12.5	3	2.75	0.89	96	681.6	63.7	90	2.25	810	26.15	360	1.99	0.2	0.2	Gen. aminoaciduria.
8.75	2.19	3.1	1.00	28	198.8	98.6	263	6.58	1214	39.2	865	4.8	0.2	0.2	Gen. aminoaciduria.
8.75	2.19	3	0.97	42	298.2	42.04	36	0.9	831	26.83	3185	17.68	0.2	0.2	Gen. aminoaciduria.
8.13	2.03	3.6	1.16	60	426	36.68	40.5	1.01	556	17.95	480	2.66	0.4	0.4	Gen. aminoaciduria.
8.75	2.19	3	0.97	60	426	63.39	108	2.7	454	14.66	3294	18.28			
mean	9.61	2.4	2.99	0.97	51.63	366.54	61.66	92.74	2.32	776.75	25.08	1326.25	7.36	0.23	
t. devia	1.53	0.38	0.34	0.11	23.7	168.7	26.95	73.81	1.85	234.5	7.51	1200.2	6.66	0.08	
St. error	0.54	0.14	0.12	0.04	8.38	59.49	9.53	26.1	0.65	82.91	2.68	424.3	2.36	0.03	

Table no. 12.

Laboratory data of the case of end organ resistance to
1,25-(OH)₂D₃ (B₆)

1,25-(OH) ₂ D ₃ 156/																	
Serum Ca		Serum P		Alk. Ph.		Cr. clear- ance		Urine Ca		Urine P		Urine glucose		Urinary ptn.		Aminoacids Chromatogram	
mg	m.mol/ L.	mg%	m.mol/ L.	K.A.U.	I.U./L.	ml./min/ 1.73m ²	mg/day	m.mol/ day	mg/day	m.mol/d.	mg./d.	m.mol/d.	gram/day				
7.5	1.88	3.4	1.1	45	319.5	82	50	1.25	267	8.62	68	0.38	0.15	Normal			

Table no. 13.

Table No. 14

Statistical significance between means of different groups

Aspect of Comparison	d.d.I.	t Value	P value	Degree of Significance
(1) <u>Serum calcium</u>				
B ₁ & A	31	4.08	< 0.001	V.H.S.
B ₂ & A	21	5.25	< 0.001	V.H.S.
B ₃ & A	17	16.80	< 0.001	V.H.S.
B ₄ & A	19	3.45	< 0.01	H.S.
B ₅ & A	21	0.62	> 0.05	N.S.
B ₆ & A	14	3.71	< 0.01	H.S.
B ₃ & B ₁	20	5.48	< 0.001	V.H.S.

D.D.I. = Degree of freedom.
 A = Control group.
 B₁ = Vit. D. deficiency group.
 B₂ = Renal Osteodystrophy.
 B₃ = Vitamin D dependent rickets.
 B₄ = Familial hypophosphatemic rickets.
 B₅ = Fanconi syndrome.
 B₆ = End organ resistance to active Vit. D.
 H.S. = Highly significant.
 V.H.S.: = Very highly significant.
 N.S. = Non significant.

cont.

Aspect of Comparison	d.d.I.	t Value	P Value	Degree of Significance
(2) <u>Serum phosphorus:</u>				
B ₁ & A	31	9.48	<0.001	V.H.S.
B ₂ & A	21	2.54	<0.02	H.S.
B ₃ & A	17	5.64	<0.001	V.H.S.
B ₄ & A	19	11.13	<0.001	V.H.S.
B ₅ & A	21	11.06	<0.001	V.H.S.
B ₆ & A	14	4.33	<0.001	V.H.S.
B ₄ & B ₁	22	5.99	<0.001	V.H.S.
B ₅ & B ₁	24	3.51	<0.01	H.S.

cont.

Aspect of Comparison	d.d.I.	t Value	P Value	Degree of Significance
(3) <u>Serum Alk. phosphatase:</u>				
B ₁ & A	31	5.59	< 0.001	V.H.S.
B ₂ & A	21	6.93	< 0.001	V.H.S.
B ₃ & A	17	15.24	< 0.001	V.H.S.
B ₄ & A	19	5.55	< 0.001	V.H.S.
B ₅ & A	21	6.6	< 0.001	V.H.S.
B ₆ & A	14	12.19	< 0.001	V.H.S.

(4) <u>Creatinine clearance:</u>				
B ₂ & A	21	18.61	< 0.001	V.H.S.
B ₅ & A	21	4.46	< 0.001	V.H.S.

cont.

Aspect of Comparison	d.d.I.	t Value	P Value	Degree of Significance
(5) <u>Urinary phosphorus</u>				
B ₁ & A	31	3.5	< 0.01	H.S.
B ₂ & A	21	3.14	< 0.01	H.S.
B ₃ & A	17	3.32	< 0.01	H.S.
B ₄ & A	19	11.91	< 0.001	V.H.S.
B ₅ & A	21	10.84	< 0.001	V.H.S.
B ₆ & A	14	3.91	< 0.01	H.S.
B ₄ & B ₁	22	10.63	< 0.001	V.H.S.
B ₅ & B ₁	24	9.58	< 0.001	V.H.S.
(6) <u>Urinary glucose:</u>				
B ₅ & A	21	4.32	< 0.001	V.H.S.
(7) <u>Urinary protein</u>				
B ₂ & A	21	2.67	< 0.02	H.S.
B ₅ & A	21	5.06	< 0.001	V.H.S.

Presentation of Some Cases

Case No 26

Vitamin D deficiency rickets

- 114 -

Case No 38
Renal Osteodystrophy

- 115 -

Case No 45

Vitamin D dependent rickets type I

- 116 -

Case No 51

Familial hypophosphatemic rickets

- 117 -

Case No 59
Fanconi Syndrome

Case No 60

End organ resistance to active vitamin D.