Summary and Conclusions

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Our study was conducted on 45 patients having late rickets i.e. rachitic patients exceeding the age of 2 years. All of our patients were picked solely on basis of clinical grounds from El-Monira University Children Hospital. Our patients included 25 males and 20 females and were ranging in age from 2 to 13 years.

Each of our patients was subjected to history taking and clinical examination with special stress on those symptoms and signs concerned with rickets and those related to each of its clinical subdivisions. Each of our patients had run the following investigations: serum calcium, serum phosphorus, serum alkakline phosphatase activity, urinary calcium, phosphorus, glucose, and proteins per 24 hours, aminoacids chromatography of the urine, and creatinine in serum and in urine from which creatinine clearance had been assessed, in addition to some selective tests directed according to underlying cause.

Our study was planned aiming at the evaluation of prevelance, various clinical and laboratory landmarks of each disorder that can contribute to the occurence of rickets beyond the age of 2 years in our children.

Throughout the study our obtained results were statistically evaluated for significance and discussed in details in the preceeding chapter. Results could be summarized into:

(1) Short stature and underweight were present in all rachitic groups but were found to be most prevelant in nonvitamin D

deficiency groups.

(2) Marfan sign was the most prevelant clinical sign for rickets met with throughout the study in every case of rickets exhibiting its manifestations before the age of 3½ years.

(3) All cases of inabelity to walk due to rickets were primary, including those cases resulting from renal osteodystrophy.

(4) Either genu varum or genu valgum is almost a constant finding in all rachitic patients with genu varum being more prevelant in all rachitic groups except for renal osteodystrophy where genu valgum was found to be more prevelant.

(5) In absence of vitamin D therapy, and as age of rachitic child advances skeletal manifestations show improvement in cases of vitamin D deficiency group, but worsens in other rachitic groups.

(6) Higher level of serum alkaline phosphatase activity in all rachitic patients than that of the control group was observed and statistically significantly higher levels in non vitamin D deficiency than in vitamin D deficiency group.

(7) Cases of vitamin D deficiency rickets were found to constitute the major bulk of late rickets below the age of 4 years, were lacking history of proper vitamin D therapy, were not associated with symptoms and signs possibly resulting from non vitamin D deficiency rickets, were having a normal or slightly lower serum calcium level, hypophosphatemia when compared to normal controls and standard values but at a degree which was found to be significantly less abnormal than Fanconi

and familial hypophosphatemic cases. Such cases also exhibited a normal urinary calcium, a normal or slightly high urinary phosphorus level, with a normal creatinine clearance and sometimes a non specific aminoaciduria. A normal response to ordinary doses of vitamin D is expected.

(8) Cases of vitamin D dependent rickets, all of whom were found to be males with history of (+) ve consanguinity, statistically significantly higher incidence of delayed walking, and hypotonia, were specifically and solely characterized by a significantly lower serum calcium level when compared to any other rachitic group (serum calcium level of 5 mg/dL.)

(9) Both cases of familial hypophosphatemic rickets and those suffering from Fanconi syndrome were characterized by statistically significant hypophosphatemia and hyperphosphaturia when compared to all other rachitic groups (serum phosphorus is usually below 3.5 mg/dL. and urinary phosphorus is usually above 0.5 gram/day). It was also noticed that cases of familial hypophosphatemic rickets, which was more prevelant in males, have exhibited minimal, if any, chest signs, frontal bossing, with a minimal degree of hyptonia which when present was concluded only from delayed walking. On the other hand, cases suffering from Fanconi syndrome were characterized by generalized aminoaciduria and glucosuria, and in some cases proteinuria and hypercalciuria. In some cases of Fanconi syndrome, creatinine clearance may be lower than normal indicating renal function impairment.

(10) Renal oesteodystrophy was found to be the leading cause of rickets above the age of 4 years. Rickets, which may be in some instances the only clinical clue to the suspect of renal failure, was usually found to present after some years of renal failure. It was found to be the only type of late rickets in which genu valgum was much more frequently met with than genu varum. Throughout the study, it was found to be the only type of rickets characterized by statistically significant hyperphosphatemia (usually more than 6 mg/dL), in addition to the lowered creatinine clearance level (statistically significantly lower than all other rachitic groups, the control group, and the standard normal values) which was found to be less than 30ml/minute/1.73 M^2 surface area in all cases of that group. In addition, some cases were exhibiting proteinuria and generalized aminoaciduria.

(11) Our single case of end organ resistance to active vitamin D shows that it can be suspected from a high age and as well, the presence of other physical stigmata characteristic of the disease as alopecia in association with significant hypocalcemia, mild to moderate hypophosphatemia and elevated serum alkaline phosphatase activity, but can be only proved by a normal serum level of 1α-25-$(OH)_2$ vitamin D_3 and lack of response to treatment with that active metabolite or its synthetic preparations in ordinary doses.

The study has thrown spotlights on some recommendations that can help, not only in the diagnostic handling of cases of

late rickets but also paving the way for further clinical and laboratory studies on late rachitic cases.

It needs not further emphasis that every clinically rachitic patient should be fully investigated not only for the sake of early detection of possible serious underlying conditions as renal failure in which renal osteodystrophy may be the only presenting feature noticed by the parents, but also in order to evaluate the required proper therapeutic regimen of vitamin D.

We find it necessary for all patients maintained on therapeutic regimens of vitamin D especially those receiving parenteral therapy to undergo serum calcium level serial determinations in order to follow up the efficacy of treatment, and as well, to ensure that patients are far from vitamin D intoxication.

We believe that a similar, but more extensive study on cases having late rickets, that includes serum levels of vitamin D active metabolites could be quiet valuable in that respect.

We also recommend further detailed studies on those cases proved to suffer from Fanconi syndrome in order to reach at an exact underlying cause in each.