# X-Linked Hypoparathyroidism : An Indian Kindred!

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## Introduction

X linked hypoparathyroidism is an extremely rare disease ,so far described in only two multigenerational kindreds in US , who later on the genetic testing were found to be interrelated. We describe X linked congenital hypoparathyroidism in a family from India.

## Case report

**Case 1** presented with hypocalcemic non-febrile generalized tonic clonic seizures at 16 months of age . Seizures controlled after hypocalcemia correction. He was put on T. calcitriol O.5mg B.D. , 1.5 g calcium /day.

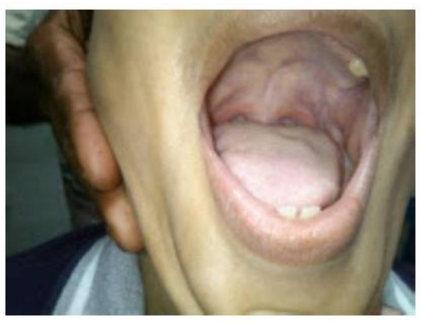
**Case 2** presented with hypocalcemic neonatal generalized tonic clonic seizure. He was put on calcitriol 0.5 mg/day , 1 g of calcium.

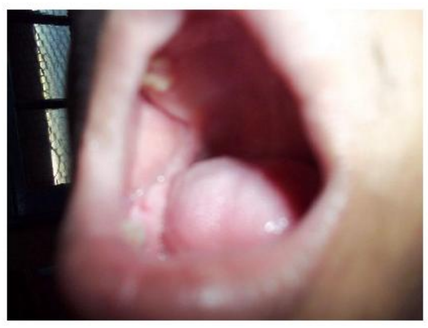
Both had delayed milestones and growth ,developed B/L cataract and B/L basal ganglia calcification, required addition of phenobabitone at 10-12 year of age.

Parents and two sisters are asymptomatic.

History of death of a male child at age of 2 years on maternal sides

Case 1(right) & case 2 (left) with orthodontia





## Investigations

| Case | S.calcium at diagnosis ( mg/dl) | Phosphorus at Diagnosis ( mmol/L) | 25(OH)Vit. D at diagnosis (ng/ml) | iPTH at diagnosis (pg/ml) | S.calcium at present (mEq/L) | Phosphorus at present (mEq/L) | iPTH at present (pg/ml) | Average 24 hr urinary calcium (mg) in last 6 years |
|---|---|---|---|---|---|---|---|---|
| 19 yr male | 1.05 | 10.61 | 16 | <1 | 8.4 | 6.1 | 5.2 | 340 |
| 14 year male | 2.3 | 9.4 | 12 | <1 | 9 | 5.3 | 3.9 | 290 |
| 20 yr old sister | 9.6 | 4 | | | | | | |
| 22 yr old sister | 9.4 | 4.3 | | | | | | |
| father | 9.8 | 3.9 | | | | | | |
| mother | 8.6 | 4.8 | | | | | | |

## Conclusion:

This is first report of X linked hypoparathyroidism outside the Missouri kindred. The two brothers exihibit strikingly similar phenotype. In addition we also describe abnormalities of dentition and cataract in presence of seemingly adequate calcitriol & calcium replacement. Genetic analysis of such family could throw insight into as yet known aspects of parathyroid gland development and disease.

## References:

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Calcium, vitamin D and bone

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