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RECURRENT ASPIRATION PNEUMONIA – A RARE PRESENTATION OF A RARE VITAMIN D DEPENDANT RICKETS TYPE1A - A CASE REPORT

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Vitamin D dependant rickets Type 1A , a rare autosomal recessive disorder [1] ,secondary to a mutation in 1α hydroxylase gene [2], which presents in early infancy with wide systemic non-specific symptoms like failure to thrive , generalised hypotonia , multiple fractures[3] , recurrent LRTIs[4] , which pose difficulty in diagnosing the ultimate endocrine defect . While some genetic disorders are devoid of specific treatment options, this disorder is fortunate to be treatable[5]. We are presenting a case of 19 months old female child, born out of a second degree consanguinous marriage, with Vitamin D Dependant rickets Type 1A, who presented with recurrent aspiration pneumonias 8 times since 8 months of her age. Her diagnosis was missed for a long time and treated at multiple places as pneumonia. She was grossly underweight, had multiple symmetrical fractures of all long bones, delayed milestones and rachitic features .Genetic analysis reported a homozygous mutation in CYP27B1 gene. She responded well to the calcitriol treatment and free of respiratory symptoms for 2 months .This multi system presentation of rickets highlights the need for high index of suspicion of possibility of rare genetic forms.

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