GROWTH HORMONE INCREASES GROWTH VELOCITY AND ALKALINE PHOSPHATASE LEVEL IN CHILDREN WITH HYPOPHOSPHATASIA

Pierre Moulin5, Jérome Sales de Gauzy2, Frédéric Vaysse3, Etienne Mornet6, Jean Pierre Salles1,4

1Endocrinologie et Maladies Osseuses, 2Chirurgie Orthopédique, 3Odontologie, Hôpital des Enfants, CHU de Toulouse, 4INSERM UMR 1043, Université Paul Sabatier, Toulouse France ; 5Service de Pédiatrie, Hôpital de Montauban, France ; 6Laboratoire SESEP, Centre Hospitalier de Versailles, Le Chesnay, France Corresponding author: Prof. J.P. Salles, Unité d’Endocrinologie et Maladies Osseuses, Hôpital des Enfants, TSA 70034, CHU de Toulouse 31059 cedex 09 France Salles.jp@chu-toulouse.fr

Hypophosphatasia is an inborn error of the metabolism, usually presenting as rickets or osteomalacia. Poor growth is a main hallmark of the disease. Growth Hormone (GH) is a powerful stimulus of growth spurt during which AP level strongly increases. We report here two cases of patients with hypophosphatasia treated with GH. Case 1: A girl was born with Intra Uterine Growth Retardation, chest and lower limbs deformity, and low levels of AP due to mutations A115V and V111M inherited from both parents. At 6 years, she presented with bone deformity, respiratory failure, dental abnormalities and severe growth delay. GH treatment was initiated at age 9 years, (50 μg/kg/week), during 6 years until final height (15 years). Case 2. A boy was seen at 9 years because of growth failure, low levels of AP associated with minimal metaphysis abnormalities. He presented with dental abnormalities, premature loss of teeth and delayed eruption of permanent teeth. Genetic analysis found two mutations, A159T and 648+1A, inherited from both parents, MRI analysis found moderate hydrocephalus, Chiari malformation and hypophysis hypoplasia. GH was started at 10 year (35 μg/kg/day), during 8 years until final height. In both cases growth velocity increased under GH, as did AP level, with no deleterious effect on the clinical bone status or mineral density parameters. Therefore, GH seems an acceptable and efficient treatment to compensate severe growth failure in hypophosphatasia.