

RICKETS AS PRECOCIOUS SIGN OF CELIAC DISEASE: THE ROLE OF VDR POLYMORPHISMS

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Vitamin D insufficiency is more frequent than expected also in Western Europe, however the relieve of a "health" child with rickets is uncommon in Caucasians.

We describe the clinical case of a 2.5-year-old girl with skeletal deformities. She was 86.5 cm (10° Cent), 12.5 kg (3-10° Cent); PH1B1.

She showed typical rickets-linked signs (costochondral swelling; Harrison's groove; genu varum; widening of wrist; skull bossing). She underwent a total-body X-ray study that showed: poor bone mineralization, femurs bowing; rachitic rosary; curved back; wrist and malleolus cupping. She had anamnestic record of insufficient sunlight exposure. She has 4 brothers with no signs of rickets.

Blood examinations revealed: Ca: 8.7 mg/dl; P: 4.7 mg/dl; Mg: 2.45 mEq/l; alkaline phosphatase: 685 IU/l; 25-OH Vitamin D: <7 ng/ml; PTH: 442 pg/ml (n.v.: 11-67); bone alkaline phosphatase: 21 (n.v.: 5-27); P1NP: 1044 (n.v.: 27-127); osteocalcin: 62.32 (n.v.: 1-11).

Deamidated Gliadin Antibodies were negative. However for the severe clinical presentation and the significant difference with the clinical condition of her brothers, she was studied for the genetic forms of vitamin D deficiency.

The VDR gene did not show any mutation; however the gene carried a homozygous transition c.2T>C, considered a polymorphism. Furthermore a heterozygous polymorphism c.1056T>C was documented in exon 9.

The sequence of CYP27B1 and PHEX genes was normal.

She received vitamin D and calcium with a significant improvement of clinical, hematological, radiographic data. However she showed a significant increase of anti-transglutaminase antibodies (IgA): 89 U/ml; (IgG): 34 U/ml (n.v.: <4); Deamidated Gliadin Antibodies (IgA): 1.8; (IgG): 26 (n.v.: <7).

The two polymorphisms (one heterozygous, associated with one homozygous) could explain the severity of rickets manifestations before celiac markers were detectable in our patient, highlighting the role of VDR polymorphisms in bone health and growth.

