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EXPERIENCES WITH MOLECULAR GENETIC INVESTIGATION OF OSTEOGENESIS IMPERFECTA IN CZECH REPUBLIC

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The incidence of osteogenesis imperfecta (OI), brittle bone disease, is 1:10-50 000 newborns. It is a heterogenous syndrome with autosomal dominant and autosomal recessive inheritance. The disease is caused by mutations of genes coding chains of collagen type I on the chromosomes 7 (Col1A2) and 17 (Col1A1). One of a few places where patients with OI are centralized in Czech Republic is The Ambulant Centre for Defects of Locomotor Apparatus in Prague. A group of 50 living patients with OI syndrome is followed up in this centre. DNA bank has been collected since the year 1998. It contains 35 isolated DNA samples. Sequenation analysis was done in 23 samples for exons 17, 27, 30, 31 of col1A1 gene and their side introns. Mutations were found in 14 patients of the studied group. Most common were substitutions in exons, substitutions in introns No. 26 and 27 were on the second place. We can´t include mutations in other exons and introns. The molecular genetic results were confronted to clinical and anthropometric findings. We verify that more mutations in one genotype area in one patient are not in correlation with clinical severity of the disease. We didn´t find any characteristics between compared patients with the same mutation. We share the idea of G. Cetta (Italy) that other genes, besides collagen genes, could interact the collagen production and become responsible for the severity of the outcome. Supported by grant of The Ministery of Education of Czech Republic EuroMISE - Cardio No. LN00B107.