We evaluated the relative frequency of each mode of inheritance in the large group of CMT fam i1ies gathered in the DNA laboratory of the Dept. of Child Neurology of the 2nd Medica!
School (including famiJies wi th already detected causal mutations). The frequency of dominant and sporadic forms is approximately equal (40% of families). In a small percentage of families (2 0k'), the autosomal recessive (AR) mode of inheritance (two or more affected siblings) was recognized. In the rest of the families (18%) there are not enough reliable data on the clinical state of the family members to indicate the mode of inheritance. Further we evaJuated the rela tive frequency of inheritance modes in the "unconfirmed" group of CMT families with ou t detected causal mutation (which previously tested negative for the CMTlA / HNPP forms and / or for mlltations in some of the following CMT-associated genes - Cx32, MPZ, PMP22, EGR2, NEFL, SIMPLE). The frequency of dominant and sporadic forms in this group is somewhat different from the large cohort of families. The frequency of dominant pedigrees is low r (30%) and the frequency of sporadic cases higher (50%). This may indicate that, in general, we can expect the detection rate of CMT causes to be higher in dominant pedigrees compared to sporadic CMT cases. In this study I have estirnated the frequency of mutations in the early growth response factor 2 gene (EGR2) among Czech patients with demyelinating form of CMT (CMTl) and with excllded most common CMT forms (CIvIT1A and HNPP). This frequency is 2,2%; (1 patient/family with detected causal mutation among 46 tested).