Genetic anticipation - true or false?

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Background: The phenomenon in which a genetic disease has an earlier onset and a more agresive evolution with each succeeding generation was controversial. However, anticipation has now been proven to occur in a large number of important genetic disorders, including myotonic dystrophies. Case report: A 41-year-old man has had locomotor difficulty since he was 18 years old. Later, he noticed difficulties in vowel pronunciation and recently, a slowed relaxation following a normal muscle contraction. Neurological examination reveal hypertonic hands and feet muscle, atrophy in the masseter, temporalis, sternocleidomastoid muscles and distal legs muscles, distal motor deficit in wrist extension,walking without aid, early balding, triangular facies. Genetic tests revealed on a chromosome (allele) 5±1 CTG repeats and on the other chromosome (allele) more than 300 CTG repeats. The pattern is characteristic for DM (muscular dystrophy) type 1 (Steinert's disease). His parents were apparently healthy, but his father has early baldness and cataracts, without any complaints. The patient has two girls, 8 and 15 years old. The older one has had signs and syptoms of DM1 disease for 4-5 years and EMG and genetic test confirmed it, with an increased number (over 300) of the CTG trinucleotide repeat in DMPK gene. Conclusion: This case report represents an exemple of a real genetic anticipation. Grandfather, father and daughter associate mild, classic and congenital phenotypes, respectively. Thus, genetic counseling and prenatal testing is mandatory for pregnancies at increased risk when the diagnosis of DM1 has been confirmed in an affected family member.