Monosomy 1p36 syndrome: reviewing the correlation between deletion sizes and phenotypes

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ABSTRACT. The major clinical features of monosomy 1p36 deletion are developmental delay and hypotonia associated with short stature and craniofacial dysmorphisms. The objective of this study was to review the cases of 1p36 deletion that was reported between 1999 and 2014, in order to identify a possible correlation between the size of the 1p36-deleted segment and the clinical phenotype of the disease. Scientific articles published in the (National Center for Biotechnology Information; NCBI http://www.ncbi.nlm.nih.gov/pubmed) and Scientific Electronic Library Online (www.scielo.com.br) databases were searched using key word combinations, such as "1p36 deletion", "monosomy 1p36 deletion", and "1p36 deletion syndrome". Articles in English or Spanish reporting the correlation between deletion sizes and the respective clinical phenotypes were retrieved, while letters, reviews, guidelines, and studies with mouse models were excluded. Among the 746 retrieved articles, only 17 (12 case
reports and 5 series of cases), comprising 29 patients (9 males and 20 females, aged 0 months (neonate) to 22 years) bearing the 1p36 deletions and whose clinical phenotypes were described, met the inclusion criteria. The genotype-phenotype correlation in monosomy 1p36 is a challenge because of the variability in the size of the deleted segment, as well as in the clinical manifestations of similar size deletions. Therefore, the severity of the clinical features was not always associated with the deletion size, possibly because of the other influences, such as stochastic factors, epigenetic events, or reduced penetration of the deleted genes.

**Key words:** Monosomy 1p36 syndrome; 1p36 deletion syndrome; Deletion size; Phenotype