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The Contribution of the PCR-Enzymatic Digestion in the Positive Diagnosis of Proximal Spinal Muscular Atrophy in the Moroccan Population

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Abstract : The proximal spinal muscular atrophy (SMA) is a group of neuromuscular disorders characterized by progressive muscle weakness due to the degeneration and loss of anterior motor neurons of the spinal cord. Depending on the age of onset of symptoms and their evolution, four types of SMA, varying in severity, result in a mutations of the SMN gene (survival of Motor neuron). We have analyzed the DNA of 295 patients referred to our genetic counseling; since January 1996 until October 2014; for suspected SMA. The homozygous deletion of exon 7 of the SMN gene was found in 133 patients; of which, 40.6% were born to consanguineous parents. In countries like Morocco, where the frequency of heterozygotes for SMA is high, genetic testing should be offered as first-line and, after careful clinical assessment, especially in newborns and infants with congenital hypotonia unexplained and prognosis compromise. The molecular diagnosis of SMA allows a quick and certainly diagnosis, provide adequate genetic counseling for families at risk and suggest, for couples who want prenatal diagnosis. The analysis of the SMN gene is a perfect example of genetic testing with an excellent cost/benefit ratio that can be of great interest in public health, especially in low-income countries. We emphasize in this work for the benefit of the generalization of molecular diagnosis of SMA by the technique of PCR-enzymatic digestion in other centers in Morocco.

Keywords : Exon7, PCR-digestion, SMA, SMN gene
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