عنوان مقاله:

Progeroid Syndrome and Mutation in LMNA Gene : Report of Two Cases from Iran

محل انتشار:

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خلاصه مقاله:

Two Iranian cases with very rare progeroid syndrome are reported. The first is a ۲۴-year-old girl who has been healthy till her ۱۳th birthday. From that time she has been suffering from a progressive generalized and multi-systemic illness. The cardinal clinical findings were growth retardation, subcutaneous fat loss, skin dryness and wrinkling, scattered focal sclerodermoid-like changes, prominence of superficial vessels, gradual loss of scalp hair and eyebrows and cardiac involvement in the form of dilated cardiomyopathy. All the above findings were suggestive of precocious ageing and the clinical diagnosis of Werner syndrome. The second case is a ۶-year-old boy with typical clinical findings of Progeria or Hutchinson-Gilford syndrome. The diagnoses were confirmed by molecular analysis of the samples in Washington and Marseille.In the first case there was no molecular abnormality in Werner's gene(WRN), but there was a mutation in the LMNA gene. The mutation was substitution of C to G in codon number ۵۷, and the codon GCA (alanine) changed to CCA (proline). So, in the codon ۵۷ of the protein Lamin A/C proline had replaced alanine (A۵۷>P). The mutation in the second case (Progeria=Hutchinson-Gilford syn.) was a point mutation at the exon ۱۱ of Lamin A/C protein resulting in the replacement of thymine by cytosine in the nucleotide number ۱۸۲۴(۱۸۲۴C>T). The importance of lamins and the mechanism and pathogenesis of progeroid syndromes are discussed briefly.

کلمات کلیدی:

Progeria, Hutchinson-Gilford syndrome, Werner's syndrome, Lamin A/C, Laminopathy, Precocious ageing