كلية الطب – جامعة الفيوم
قسم الأطفال

البحث الثامن

عنوان البحث:

Clinical and Genetic Characterization of Ten Egyptian Patients with Wolf-Hirschhorn Syndrome and Review of Literature

مكان وتاريخ النشر:
Molecular Genetics&Genomic Medicine. October 2020;00:e1546. Pages 1-17

Abstract

Wolf–Hirschhorn syndrome (WHS) (OMIM 194190) is a multiple congenital anomalies/intellectual disability syndrome. It is caused by partial loss of genetic material from the distal portion of the short arm of chromosome. **Methods:** We studied the phenotype–genotype correlation. **Results**: We present the clinical manifestations and cytogenetic results of 10 unrelated Egyptian patients with 4p deletions. Karyotyping, FISH and MLPA was performed for screening for microdeletion syndromes. Array CGH was done for two patients. All patients exhibited the cardinal clinical manifestation of WHS. FISH proved deletion of the specific WHS locus in all patients. MLPA detected microdeletion of the specific locus in two patients with normal karyotypes, while array CGH, performed for two patients, has delineated the extent of the deleted segments and the involved genes. LETM1, the main candidate gene for the seizure phenotype, was found deleted in the two patients tested by array CGH; nevertheless, one of them did not manifest seizures. **Conclusion**: WHS is a contiguous gene syndrome resulting from hemizygosity of the terminal 2 Mb of 4p16.3 region. The Branchial fistula, detected in one of our patients is a new finding that, to our knowledge, was not reported.