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A new case of de novo chromosome 19p13.12 deletion in an Omani girl with global developmental delay and multiple congenital anomalies**Musallam Al Araiimi, Salma Al Harasi, Nishath Hamza, Manal Al Kharusi, Hibah Al Hasni, Ali Al Yahmadi and Waad Allah Mula Abed**
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1 9p13.12 deletion syndrome is a rare genetic disorder in which a small section of the short arm of chromosome 19 is deleted. It is a newly identified syndrome which is characterized by developmental delay, learning impairment and facial dysmorphism. We report a 4-year-old Omani girl with 19p13.12 micro-deletion syndrome. She was born as full-term to a non-related parent with global developmental delay, hypotonia and dysmorphism. She presented with multiple phenotypic skeletal abnormalities, hypotonia, and facial dysmorphism including frontal bossing, down slanting palpebral fissures, maxillary hypoplasia, bi-temporal narrowing, arachnodactyly and strabismus. Skeletal survey radiographs revealed thin long bones and square shaped of some vertebral bodies. Computed tomography (CT) and Magnetic Resonance Imaging (MRI) of the brain were unremarkable. Parents and the older sibling daughter were asymptomatic. Using array comparative genomic hybridization (CGH) analysis, a novel 1,594 kbp deletion at 19p13.12 was identified with 53 genes on which 35 are OMIM genes. These include *NFIX* (OMIM #164005), *MAN2B1* (OMIM #609458), *NFIX* (OMIM # 615094), *CACNA1A* (OMIM # 601011) and *GCDH* (OMIM # 608801) that could be responsible for the presented phenotypes (global developmental delay and various skeletal anomalies). This was found to be a de novo mutation by investigating the parents. We present this patient as the first case reported in Oman and the Gulf region.

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