

Oral and dental manifestations in KBG syndrome

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KBG syndrome is an intellectual disability (ID) associated with multiple congenital anomalies in which the macrodontia could be the clue for the diagnosis. It is caused either by heterozygous variant in *ANKRD11* gene or 16q24.3 microdeletions. Here, we report on two unrelated male patients presented with ID, short stature, webbing of neck, and cryptorchidism. Noonan syndrome was suspected first but the presence of macrodontia in both patients pointed out to KBG syndrome which was confirmed thereafter by the identification of a novel pathogenic variant in *ANKRD11* gene, c.5488G>T (p.E1830*). Macrodontia was noticed in all the deciduous anterior teeth in Patient 1. This observation was reported previously in few patients, but it seems to be a common feature that could be misdiagnosed as premature eruption of teeth. Therefore, our results confirm that maxillary permanent central incisors may not be the only teeth affected in KBG but also all the deciduous. Interestingly, desquamative gingivitis was additionally noted in Patient 1, which has not been reported previously. To the best of our knowledge, this is the first report from Egypt.

Biography

Dr. Sayed obtained Bachelor degree, from faculty of Oral and dental Medicine, Cairo university with the degree of very good (honor). This gave her the opportunity to get a position in the Oro-dental Genetics Department in National Research Centre in 2001. In 2007, she was awarded M.Sc degree in Oral Biology and in 2015, she was awarded Ph.D. degree in dental laser applications from Cairo University. Her duties in Oro-dental Genetics Department include oro-dental examination of patients attending Orodental Genetics clinic, Neurogentics clinic as well as Limb malformation and skeletal dysplasia clinic which aids in their diagnoses.

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