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Autor(en) des Beitrags:
Salem, Nabeel J M; Hempel, Maja; Heiliger, Katrin-Janine; Hosie, Stuart; Meitinger, Thomas; Oexle, Konrad

Titel des Beitrags:
Anal atresia, coloboma, microphthalmia, and nasal skin tag in a female patient with 3.5 Mb deletion of 3q26 encompassing SOX2.

Abstract:
A full term female newborn presented with prominent forehead, bilateral microphthalmia, iris coloboma and cataract, wide intercanthal distance, large, low-set and protruding ears, skin tag at the left nasal nostril, imperforate anus with rectovestibular fistula, and postnatal growth delay with brachymicrocephaly. A marker chromosome was not detectable and the copy number of 22q11 was normal. However, array CGH revealed a 3.5 Mb microdeletion of chromosome region 3q26.32-3q26.33 (chr. 3: 178,598,162-182,114,483; hg19) which comprised the SOX2 gene. While SOX2 haploinsufficiency is known to cause microphthalmia and coloboma, it has not been described before in patients with anal atresia.

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