Iris mammillations in two female siblings with congenital adrenal hyperplasia.

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Source

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Abstract

Iris mammillations are dark brown, smooth, mound- or dome-shaped protuberances that are typically found on the anterior iris surface and are presumed to be congenital in origin. This congenital anomaly is usually unilateral and can be hereditary or sporadic. Lisch nodules in neurofibromatosis, tapioca melanoma of the iris, inflammatory iris granulomata and Cogan-Reese syndrome should be considered in the differential diagnosis. In this case report, the authors present a case of a bilateral iris mammillations in two siblings with congenital adrenal hyperplasia (CAH). To our knowledge, this is the first case where bilateral iris mammillations have been found to be associated with a systemic condition. Iris mammillations can be considered as one of the clinical signs in CAH in view of the pathogenesis discussed. Detailed ocular examination in CAH may reveal an increased incidence.