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A case report of optic disk drusen in the patient with partial trisomy 16q

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Chromosome 16 abnormalities cause a wide spectrum of disorders associated with a variety of outcomes. Full trisomy 16 is the most frequent autosomal cause of miscarriages due to associated malformations. While complete trisomy 16 can be considered to be lethal in early pregnancy, mosaic trisomy 16 have been reported. Partial trisomy 16 for the entire long arm is rare-Partial trisomy 16q is regarded as a clinically recognizable entity presenting with intrauterine growth restrinction, dysmorphic features, central nervous system malformations, generalized hypotonia, absence of suck, severe psychomotor retardation and intellectual disability, congenital heart defects, ambiguous genitalia, vertebral and anorectal anomalies. Several ocular abnormalities have been described in partial trisomy 16q patients, such as bilateral anterior segment dysgenesis with iris hypoplasia, Rieger's anomaly, congenital glaucoma and megalocornea. In this paper we report our experience following a female patient with partial trisomy 16q. Her clinical features appeared similar to those reported in the literature (low birth weight, microcephaly, congenital heart defect, pre and postnatal growth and mental retardation, seizures, mild dysmorphic features, liver and central nervous system anomalies). However, she also developed a bilateral optic nerve head drusen, confirmed by OCT that has never been described in literature.

Biography

Morreale Bubella Raffaella is a Specialist in Ophthalmology and has obtained PhD in Neuroscience and disorders of comportment. She has published more than 36 papers in reputed journals and a book *"L'occhio nella patologia endocrina"*.

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