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Case report

Achiasma and Kapur-Toriello syndrome: Two rare entities

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ABSTRACT

Purpose: Describe a unique case of achiasma in a patient with Kapur-Toriello syndrome.
Observations: We provide a brief review of achiasma with common findings on examination and imaging studies, and a published classification system. In addition, consistent with the rare Kapur-Toriello syndrome, he had a right unilateral cleft lip and palate, neurologic abnormality in his achiasma, anal atresia, vesicoureteral reflux, hypospadias, and growth deficiency.
Conclusions: and Importance: Achiasma is an incredibly rare neurological deficit and we present the first case of achiasma in Kapur-Toriello syndrome.

1. Introduction

We describe a case of achiasma in a 5-year-old male with a constellation of signs demonstrating likely Kapur-Toriello syndrome. Our paper reviews achiasma, discusses a published classification system, and studies the common findings on examination and imaging studies. In addition, our patient represents on the seventh patient described with Kapur-Toriello syndrome, as our patient had a right unilateral cleft lip and palate, neurologic abnormality in his achiasma, anal atresia, vesicoureteral reflux, hypospadias, and growth deficiency. He represents the first case of achiasma in Kapur-Toriello syndrome.

2. Case report

A 5-year-old Caucasian male presented at Wilford Hall Ambulatory Surgical Center for evaluation by pediatric ophthalmology after achiasma was noted on MRI. The MRI was ordered by his pediatric endocrinologist as part of his evaluation for his growth deficiency. He had a history of right unilateral cleft lip and palate that had undergone repair and rhinoplasty, anal atresia, vesicoureteral reflux, hypospadias, 11 ribs, growth hormone deficiency, hypothyroidism, pes planus and transsphenoidal encephalocele. He was evaluated a pediatric geneticist who felt that his constellation of symptoms were consistent with Kapur-Toriello syndrome. No history of birth defects, severe cognitive impairment, genetic conditions, recurrent miscarriages or sudden death were noted in his family. He had been previously evaluated by pediatric ophthalmology when he was 3 months and at that time was noted to have an iris coloboma OD, a retinochoroidal coloboma OD, and a horizontal nystagmus.

Ophthalmic examination found a visual acuity of 20/250 OD and 20/150 OS. Extraocular movements were restricted OD with upshoot in left gaze consistent with either an intranuclear ophthalmoplegia or Duane's syndrome (Fig. 1). There was no abnormality of the VIth nerve noted on MRI. He had a right relative afferent pupillary defect. He had a right exotropia 35 PD at near and 30 PD at distance (Krimsky). In addition, he had a small amplitude, high frequency pendular horizontal nystagmus with overlying right beating nystagmus (Supplementary Video 1). On slit lamp examination, he had an inferior right iris coloboma. His posterior examination demonstrated a large retinal and optic nerve coloboma (Fig. 2A). The rest of his OD and his entire OS anterior and posterior examination was normal.

Supplementary video related to this article can be found at https://doi.org/10.1016/j.ajoc.2019.100488.

Chromosome microarray showed a small gene deletion of 6p23, which is not associated with any syndrome. Kapur-Toriello syndrome is not associated with any gene abnormalities. MRI demonstrated hypoplasia of both optic nerves and achiasma (Fig. 2B and C,D). In addition, a sphenonasopharyngeal/transphenoid basal cephalocele involving the meninges and sellar contents and thin flattened enhancing pituitary tissue along the floor and anterior margin of the cephalocele were noted. Both Humphrey and Goldman visual field testing were attempted, however due to the patients age, reliable testing was not possible.

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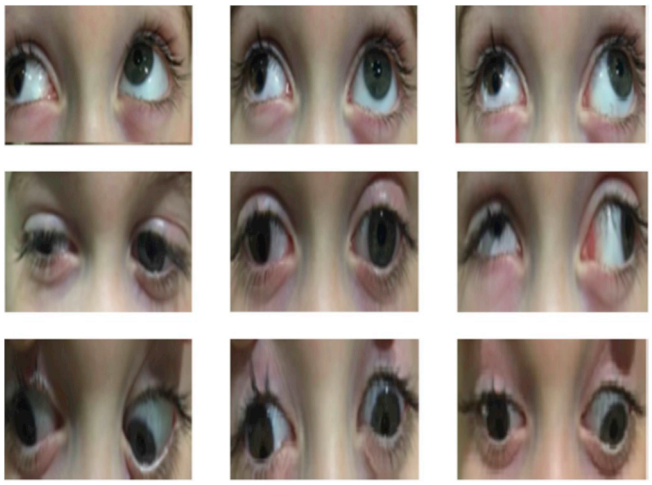

Fig. 1. Extra-ocular movements with restriction of the right eye on left gaze with upshot.

3. Discussion

Achiasma is an uncommon condition consisting of a lack of an optic chiasma with each optic nerve supplying only its ipsilateral visual cortex. Non-decussating retinal fugal fiber syndrome[1] has historically been described with very specific features of a combined horizontal and seesaw nystagmus, achiasmic VEP pattern, normal visual field and foveal reflexes, and normal endocrine function. However, another case series[2] recognized that most patients do not fall within this criteria and developed a separate classification system: Type A as patients with reduced decussation with optic nerves of normal appearance on clinical examination, type B as those with reduced decussation, chiasmal hypoplasia and midline defects of septo-optic dysplasia (absent septum pellucidium, optic nerve hypoplasia and hypopituitarism), and type C as those with reduced decussation, chiasmal hypoplasia and clefting disorders and encephaloceles. Our patient appears to fall in both type B and C.

Visual field testing is surprisingly generally normal. One study[3] of interest evaluated the visual system in two achiasmic patients. These patients had normal visual field sensitivities and no visual field defects. They found overlapping visual hemifield maps. In addition, the achiasmic patients had normal geniculate-cortical and occipital-callosal connections, and relatively unaltered cortico-cortical connections. Another paper,[4] further evaluated and found the hemifields columns co-opted the ocular dominance columns in patients with human achiasma.

Finally, our patient had signs consistent with a diagnosis of Kapur-Toriello syndrome. Kapur-Toriello syndrome is a very rare multiple congenital anomaly syndrome[5,6,7] with only 6 cases described to date. The constellation of features includes cleft lip and palate, heart defects, intestinal abnormalities, a distinctive nose with a bulbous tip and long columella below the nares, and low set ears. In addition, growth retardation, microphthalmia/anophthalmia, coloboma, digital abnormalities, cognitive impairment, and neurological abnormailities are variably noted. On initial evaluation, our patient had growth retardation related to growth hormone deficiency and hypothyroidism,

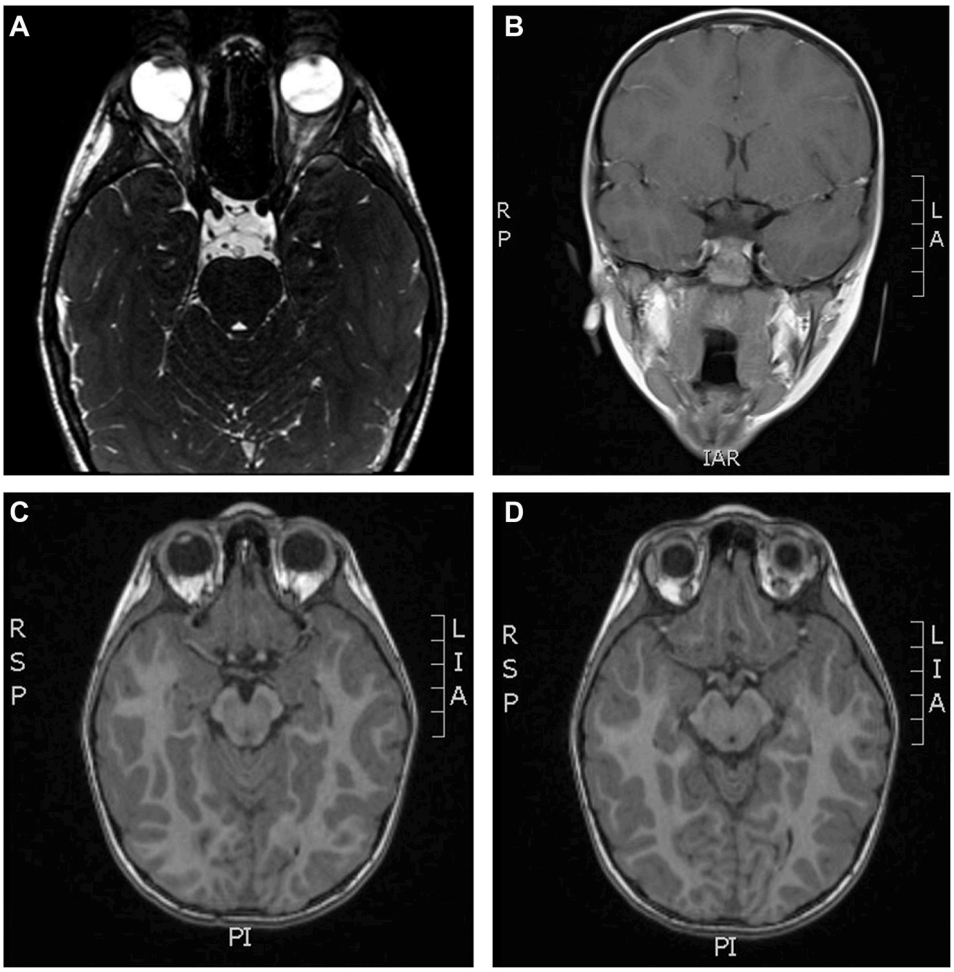

Fig. 2. MRI a. Optic Nerve Coloboma (axial cut), b. Coronal cut demonstrating achiasma above pituitary gland. C,D Axial cuts demonstrating achiasma.

unilateral cleft lip and palate, iris and optic nerve coloboma, and anal atresia. However, although he does not have the low set ears, he does have overfolding and posteriorly rotated ears. In addition, his nose appears bulbous, but he does not appear to have an overextended columella as described in the 6 previous cases. It is possible that the overextended columella was fixed during his rhinoplasty. Previously a wide range of neurologic abnormalities have been noted in Kapur-Toriello syndrome, including midline deficits like corpus callosum and pituitary abnormalities, white matter atrophy, and Dandy-Walker malformation.[6] This is the first case in which achiasma was the noted neurologic abnormality, although as discussed above midline deficits have been noted. Per evaluation by the pediatric geneticists, his spectrum of signs were most consistent with a diagnosis of Kapur-Toriello syndrome although he did consider possible diagnoses of CHARGE, Opitz G/BBB and Kabuki syndrome. This differential is consistent with that considered by other authors.[6]

In conclusion, achiasma and Kapur-Toriello syndrome are both rare congenital anomalies. This manuscript represents the first case report of achiasma in a patient with Kapur-Toriello syndrome.

Patient consent

Written consent to publish case details and photographs of the patient were obtained from the patient's parents/legal guardians.

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Conflicts of interest

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Disclaimer

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Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.ajoc.2019.100488.

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