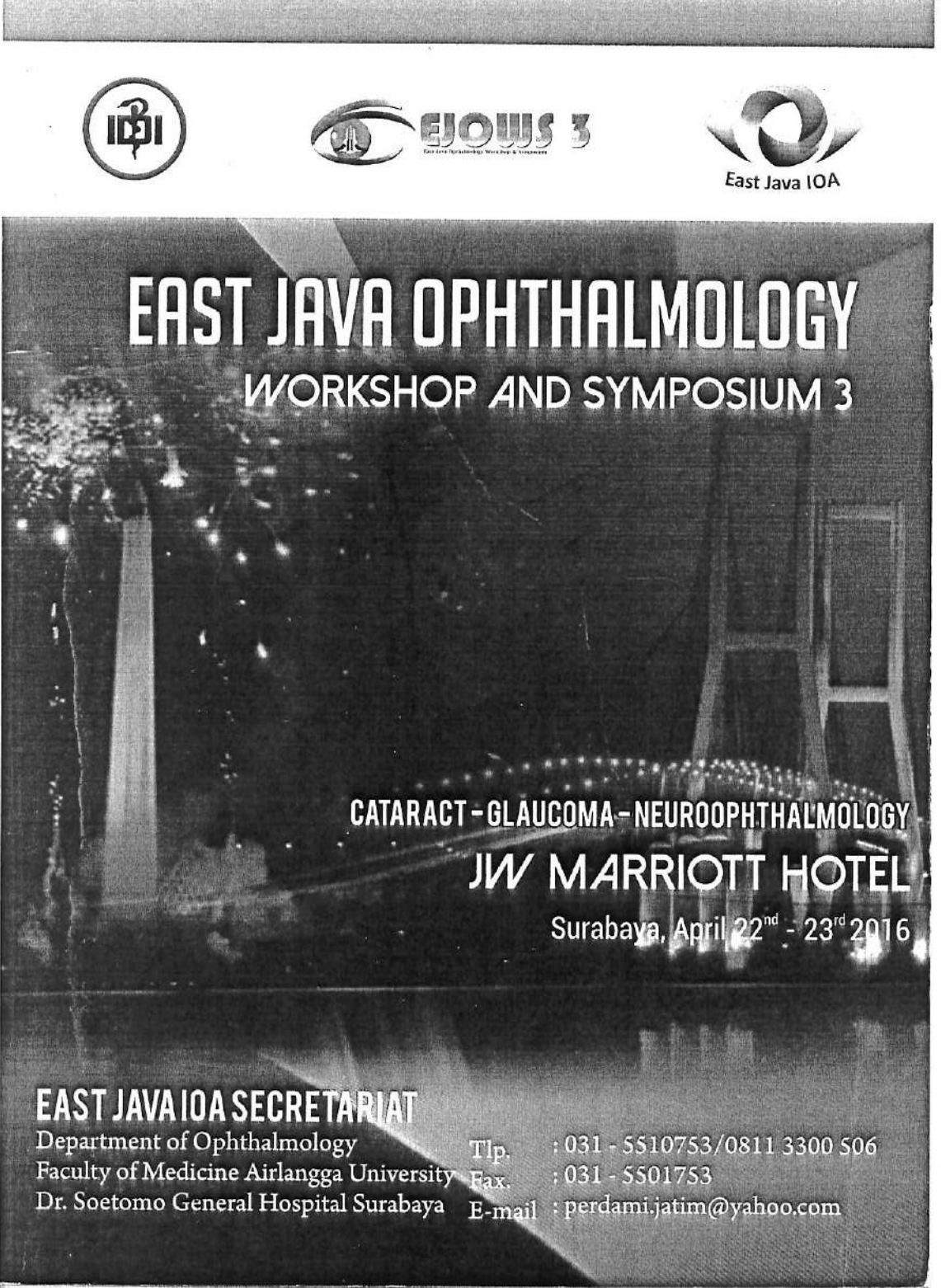
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Optic Nerve Hypoplasia in a Child with Midline Cerebral Structures Anomalies

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Introduction

Optic nerve hypoplasia (ONH) is a congenital abnormality characterized by small optic discs affecting one or both eyes. It can occur in isolation or in combination with a myriad of functional and anatomic abnormalities of the central nervous system. Recently, ONH has been recognized as an increasingly frequent problem. Concomitantly, our understanding of this epidemic of neuronal dysgenesis and its diverse impact on growth and development has evolved considerably.[1]

The Case

A, 6 years-old boy admitted to the hospital with a chief complaint of not having eyes contact since 6 month of age. Since 40 days old, his parents realized that both of his eyes were moving in circular motion and aren't able to fixate on (or follow) a certain object until this moment. The boy has no history of seizure before. The patient was able to position himself in a prone (crawling) at the age of 6 months, stand at the age of 2 years old and walk at the age of 3. He is able to speak individual words with limited vocabulary (with the tendency of repeating/copying others) but not yet able to form sentences. No one in the family has a similar medical condition. Patient was the 6th of eight children in the family. History of domestic violence during pregnancy was found.

Patient had a history of spontaneous delivery at sufficient age immediately cried after birth with a birth weight of 2800 grams. He had a history of complete immunization until 2 years old. He was also

found to be given formulated milk without breast milk.The child swallows food without chewing, is already able to grasp a certain object with assistance and is found to be able to see (identify) large objects.

The Brain MRI showed agenesis of corpus callosum, hypoplasia of optic nerve and absence of septum pellucidum.

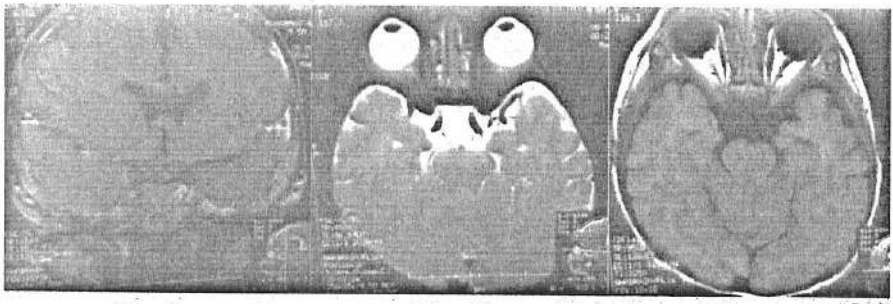

Fig 1a Fig 1b Fig 1c

Figure 1 a,b,c. MRI showed agenesis of corpus callosum, absence of septum pellucidum and optic nerve hypoplasia.

The child was diagnosed as optic nerve hypoplasia with midline cerebral structures anomalies and corservative treatment was implemented.

Discussion

Reeves first described a septo-optic dysplasia syn-drome in 1941. However, it was not until 15 years later that de Morsier coined the expression septo-optic dys-plasia (SOD), characterized by agenesis of the septum pel-lucidum and optic nerve hypoplasia . Another 15 years passed before Kaplan et al. demonstrated that this syndrome is, to a high degree, associated with pitu-itary hormone insufficiencies. Children with optic nerve hypopla-sia and SOD have been reported to be blind in 25±50%, or severely visually impaired in

another 40% [13±15]. A spe-cific ocular fundus appearance with a markedly small optic disc and isolated tortuosity of the retinal veins. Optic disc dys-plasia and morning glory sign have additionally been described in association with endocrinopathy, indicating that children with these ophthalmological abnormalities should undergo endocrinological investigation. Seizures have been reported in 25±30% and psychomotor retardation in about 50±60% of these children. Stereotypical behavioural abnormal-ities have been noted in a surprisingly high proportion (150%) of our patients.[2,3]

In our case the child suffered from severely impaired of vision since he was 6 months of age. Unfortunately the patients could not afford to perform endocrinological testing. The MRI showed the of agenesis of septum pellucidum and optic nerve hypoplasia. There was neurological disturbances and physically mentally retarded, otherwise no seizure was observed. In order to have a proper and more specific diagnosis, a child should be investigated as suggested above. Thus, one should describe abnormalities of: (1) the hypothalamo-pituitary axis, including the extent of endocrinopathy; (2) the anterior visual pathways (optic nerve hypoplasia, coloboma or dysplasia), including the extent of visual impairment, and (3) other midline brain structures (e.g., septum pellucidum, corpus callosum). In order to have a more holistic and functional diagno-sis, the degree of neurological, neuropsychiatrical and psychological involvement should also be stated in each case.[1,2,4]

Occupational, physical, and/or speech therapy are frequently needed by children with ONH. Attention should especially be given to early development of oral motor skills and acclimation to textured foods for those children resistant to eating. Incorporating words into song can sometimes ameliorate delayed verbal communication.

The vision of young children with ONH should be mon-itored at least annually, and any refractive errors should be treated when

the visual acuity reaches a functional level. Patching of the better eye can result in improvement of vision in the worse eye. However, if the ONH is asym-metric, maintenance of improved vision requires prolonged patching that can be disruptive to development in a child with many other handicaps. Thus, amblyopia therapy should be reserved for those cases in which the potential vision in each eye is felt to be fairly good.[1,2]

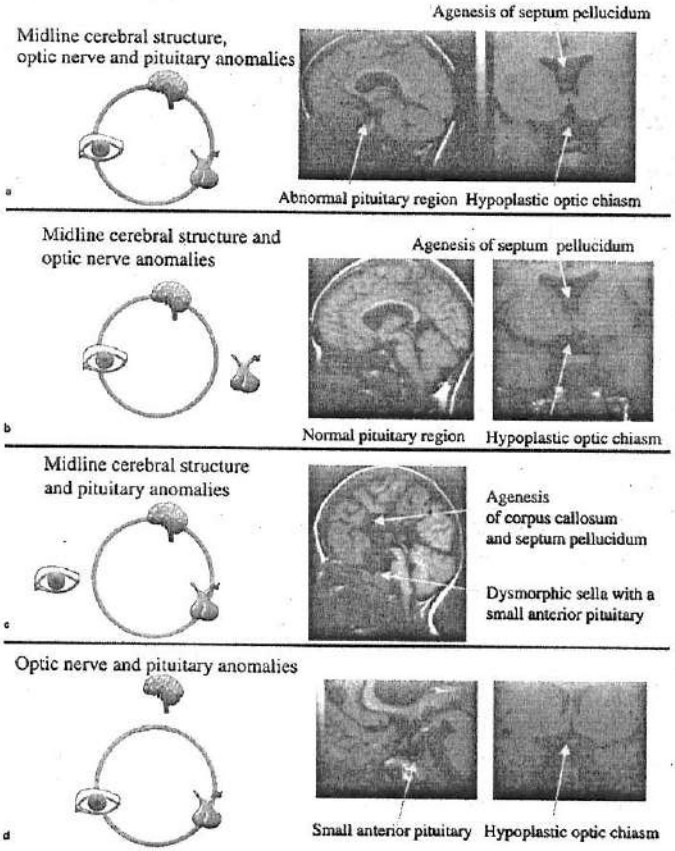

Fig 2. Varies of optic nerve hypoplasia syndrome[2]

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