Septo-optic dysplasia

Septo-optic dysplasia (SOD), (de Morsier syndrome) is a rare congenital malformation syndrome featuring underdevelopment of the optic nerve, pituitary dysfunction, and absence of the septum pellucidum (a midline part of the brain). Two of these features need to be present for a clinical diagnosis — only 30% of patients have all three [1) 2)].

Georges de Morsier first recognized the relation of a rudimentary or absent septum pellucidum with hypoplasia of the optic nerves and chiasm in 1956 [3)].

The frequently associated features of hypopituitarism and absent septum pellucidum were felt to have embryonic linkage as “septo-optic dysplasia” or “de Morsier's syndrome.” More recent studies have suggested these associations are independent of one another [4)].

Incomplete early morphogenesis of anterior midline structures produces hypoplasia of the optic nerves and possibly optic chiasm (affected patients are blind) and pituitary infundibulum. The septum pellucidum is absent in about half the cases. About half the cases also have schizencephaly.

The presentation of Septo-optic dysplasia may be due to secondary hypopituitarism manifesting as dwarfism, isolated growth hormone deficiency, or panhypopituitarism. Occasionally hypersecretion of growth hormone, corticotropin or prolactin may occur, and precocious puberty may occur. Most patients are of normal intelligence although retardation may occur. Septo-optic dysplasia may be a less severe form of holoprosencephaly, and occasionally may occur as part of this anomaly (with its attendant poorer prognosis for function or survival). The ventricles may be normal or dilated. May be seen by the neurosurgeon because of concerns of possible hydrocephalus.

Case series

Sixty-four patients with SOD were identified, and 7 of those patients (10.9%) were diagnosed with hearing loss. Type of hearing loss was sensorineural (SNHL) in 5 patients (63%), mixed (MHL) in 1(14%), and conductive (CHL) in 1(14%). Bilateral loss presented in 60% (3/5) of SNHL patients, while the rest demonstrated unilateral loss. Unilateral findings included cochlear nerve deficiency (1) and atresia/microtia (1). Tympanostomy tubes were required in 57% (4/7) of SOD children with hearing loss. Amplification was successfully implemented in 86% (6/7).

Hearing loss was found in nearly 11% of SOD children, and SNHL was identified as (63%) the predominant form of loss. To our knowledge, this is the first retrospective review of hearing loss in a pediatric SOD cohort and the first to report of cochlear nerve deficiency and atresia/microtia in this population. Based on these findings, early identification of hearing loss with imaging when appropriate and treatment of otitis in this population is recommended [5)].

1)
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