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Calloso-septo-optic dysplasia-plus (De Morsier's syndrome) with aqueductal stenosis and posterior cervical myelomeningocele: Magnetic resonance imaging: A case report/series of De Morsier's syndrome

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Introduction: Septo-optic dysplasia (SOD) or De Morsier's syndrome is diagnosed when optic nerve hypoplasia is seen in conjunction with dysgenesis of the septum pellucidum. Its diagnosis is mainly neuroradiologic with an incidence of 1:50,000. This paper presents four cases of SOD diagnosed via magnetic resonance imaging in less than a year in our institution. The cases represent the classic SOD as well as an unusual blend of the neuroradiologic features of the different SOD subsets with unique associations (aqueductal stenosis and posterior cervical myelomeningocele).

Case Presentation: 1) An 18-month Filipino female with seizure, developmental delay and hydrocephalus revealed an absent septum pellucidum and corpus callosum, small optic nerves, dilated ventricles, aqueductal stenosis, grey matter heterotopia and a posterior cervical myelomeningocele; 2) A 15-year old male with recurrent seizure showed dysplastic optic nerves, dilated ventricles with box-shaped frontal horns, absent septum pellucidum and a thinned-out corpus callosum; 3) A 17-month old male with blurred vision demonstrated cerebral atrophy, open-lip schizencephaly, absent septum pellucidum with dilated ventricles and atrophic optic nerves; 4) A three-week old male with seizures since birth showed absent septum pellucidum with box-like appearance of the

frontal horns of the lateral ventricles.

Conclusion: Coincidence of seizures, developmental delay, calloso-septo-optic-dysplasia plus, aqueductal stenosis, hydrocephalus and cervical myelomeningocele is a unique constellation of the neuroradiologic features of the different subsets of SOD with indefinite prognosis. Patients with classic SOD or calloso-septo-optic dysplasia plus with rare associations should both be closely followed up for re-assessment, further evaluation and management of neurologic and non-neurologic deficits.

Speaker Biography

Joy A David has completed her Doctor of Medicine from the University of Santo Tomas, Philippines. She has worked in the field of Public and Community Health for nine years before she entered Residency Training in Radiology. She is in her second year of training at Bicol Medical Center (BMC) Naga City, Camarines Sur, Philippines and is currently the Chief Resident of the BMC Department of Radiology. She has presented her case paper in both local and international gathering/congress. She is also a Certified Women and Child Protection Specialist.

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