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Updated diagnostic criteriae defining ‘CHARGE’ Syndrome: A case scenario

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Introduction: Pagon et al coined the term ‘CHARGE’ an acronym to 5 major features. Restricted criteria of Pagon and Blake et al, dichotomously divided, made diagnosis of this entity rare, giving rise to ‘possibly’ CHARGE or ‘Near’ CHARGE like terms. Major Signs of CHARGE Syndrome are (3C): Coloboma(iris/choroid, with or without microphthalmia, Atresia of choanae, Hypoplastic semi-circular Canals. The Minor Signs of CHARGE Syndrome are, Rhombencephalic dysfunction(brainstem dysfunctions, cranial nerve VIIto XII palsies and neurosensory deafness), Hypothalamo-hypophyseal dysfunction(including GH and gonadotrophin deficiencies), Abnormal middle or external ear, Malformation of mediastinal organs(heart,esophagus) and Mental retardation.

Case Discussion: Term, AGA, Male baby, 4th order birth to Non-consanguinously married couple, delivered by normal vaginal route, through meconium stained amniotic fluid, birth weight being 3.2 kgs. Mild respiratory distress since birth required supplemental oxygen and referred to us on D4. On admission, baby had significant respiratory distress, Downe's score being 6/10, taken on conventional mechanical ventilation for 48 hours, weaned to hood. Dysmorphic features observed were bilateral nanophthalmos, high and narrow arched palate, Lt.choanal atresia. Chest Xray showed fluffy opacities in bilateral lungs, Echo screening showed 2 ASDs, 1 VSD, USG abdomen revealed Rt Renal agenesis, MRI Brain being normal, OAE b/l ear-Refer. Ophthalmologist opined very guarded prognosis of vision.

Discussion: CHARGE is characterized by very specific developmental anomalies of optic vesicle, otic capsule, midline CNS structures and upper pharynx. Updated definition of CHARGE reinforces very specific embryological defects avoiding inclusion of nonspecific or secondary anomalies and dismiss sex-dependent criteria. 3 Major criteriae refer to 3 non-overlapping embryonic territories, 5 Minor criteriae to topographically distinct areas. Hence 3 major criteria fulfilled in our case study, defines the syndrome.

Biography

Laxmi S Hadalagi, after completing MBBS in the year 2002 at my hometown, she perceived paediatrics as she was very much interested in this field and completed my postgraduation in reputed medical college of Karnataka i.e Bapuji and Chigateri Government Hospital, Davangere, Karnataka. Seeing many neonates suffering from birth asphyxia, she always questioned herself, Is there no treatment available to prevent damage of these growing brains???This zeal took her towards doing Fellowship in Neonatal and Perinatal Medicine and started working at a Tertiary Care Unit catering to peripheries, started Therapeutic Hypothermia with Cerebral Function Monitoring and Erythropoietin to reduce neuronal damage. This has given them satisfactory results in terms of mortality and neurodevelopmental outcome. She had been to Georgetown University Hospital, Washington DC,2015, as GIANI Scholar Awardee(By AAP and NNF), appreciating her research work and community level works. She is now working as Unit Incharge, examiner for Fellowshipexam for Doctors and Nurses in Neonatology, also Board of Advisor for Public-Private Partnership(VIJINAP 2016) to upgrade our government hospitals to meeting health demands of our society. Conduct regular workshops/CME for doctors and nurses to keep them updated.

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