

CASE REPORT

MICROCEPHALY WITH HYDRANENCEPHALY SYNDROME: RARE CASE REPORT

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CASE SUMMARY: Seven months old female infant, second born to non-consanguineous parentage presented with inability to recognize mother, lack of social smile, delayed attainment of head control. There was uneventful antenatal history. It was a full term vaginal delivery of baby weighing 2.5 kgs and cried immediately after birth.

Vitals were stable. Head circumference was 33cms, length was 57cms, and weight was 5.5kgs. Receding forehead, prominent metopic sutures, closed anterior fontanel, low set ears, long philtrum and high arched palate. (Figure A) Neurological examination revealed global delay in milestones with spasticity of all four limbs with exaggerated reflexes and extensor plantar response. Other systems were normal clinically. Hemoglobin was 11.2gm/dl, total count was 11,600/mm³. IgM and IgG antibodies for TORCH infection were negative. Karyotyping revealed 46, XX chromosomes. T1W and T2W Magnetic resonance imaging of Brain showed severe thinning of supra tentorial parenchyma with relative sparing of thalami and basal ganglia, falx is maintained. The lateral and third ventricles are dilated with disproportionate dilation of temporo occipital horns. Corpus callosum thinning is also noticed (Figure B, C, and D). There was evidence of tonsillar herniation and indentation of posterior craniocervical cord. All these features were suggestive of hydranencephaly with hydrocephalus. Counseling was done regarding prognosis of condition. Mother was trained about feeding, physiotherapy.

DISCUSSION: Microcephaly is defined as head circumference more than -3SD below for that age and sex.^[1] Hydranencephaly is near total absence of brain parenchyma with intact meninges and cranial vault. Basal ganglia are also usually affected. Association of microcephaly with hydranencephaly called micro hydranencephaly is uncommon.^[1] Possible hypothesis include massive infarction secondary to obstruction in internal carotid arteries in intra uterine life.^[2] Condition is mostly recognized at birth.^[1] There has been report of syphilis, toxoplasmosis, herpes simplex, infectious hepatitis, listeriosis, trauma, ionizing radiation, rubella.^[2,3,4] Patients present with extreme microcephaly, profound motor and mental retardation, spasticity, failure to thrive, growth retardation and incomplete cerebral formation.^[5] Radiologic studies showed gross dilation of the ventricles resulting from the absence of cerebral hemispheres or severe delay in their development, as well as hypoplasia of the corpus callosum, cerebellum, and brainstem. Survival despite of near total absence of cerebral parenchyma is due to presence of brain stem. The Novel Genetic Disorder Micro hydranencephaly Maps to Chromosome 16p13.3-12.1 has been identified in a large consanguineous Anatolian family with children having microhydranencephaly and intragenetic homozygous deletion in NDE1 gene has been identified although functional studies were not performed.^[1,6] There have been attempts to diagnose the disorder prenatally by use of

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serial ultrasonographic investigation, and the fetus was reported to be normal at 8, 14, and 19 wk of pregnancy.^[1] Further evidence was needed in respect of antenatal diagnosis and interventions. Treatment is supportive.

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Fig. 1

Fig. A: Microcephaly with facial features.

Fig. B: MRI of brain (T1W) in coronal section showing thinned out cortex, preserved falx, intact thalamus and basal ganglia, ventricular enlargement.

Fig. C and D: MRI of brain (T2W) in transverse section showing thinned out cortex, preserved falx, intact thalamus and basal ganglia, ventricular enlargement.

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