

Short Communication

Delleman Syndrome: a Known Case with an Unusual Finding

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ABSTRACT

A new case of Delleman syndrome (oculocerebrocutaneous syndrome) with predominant and typical

cutaneous involvement is association with coarctation of the aorta is reported.

KEYWORDS: absence of corpus callosum, coarctation of the aorta, Delleman syndrome

CASE REPORT

M.A was a 2.750 gm boy, born at term to a 24-years old primi gravida. He is the first baby of his parents and was delivered by emergency caesarean section for reduced fetal movement. Antenatal ultrasound revealed oligohydramnios, hydrocephalus and IUGR. He was found to have multiple congenital abnormalities consisting of large head, frontal bossing, low setting ears, bilateral microphthalmia, sclerocornea with right eyelid skin tag, bilateral eyes coloboma and skin hypoplasia of the scalp. He was admitted to the neonatal intensive care unit for further evaluation. Soon after admission the baby developed generalized tonic-clonic convulsions, which proved to be epileptiform runs over the left mid-temporal region on electroencephalogram requiring a loading dose of phenobarbitone to abort the seizures. Urgent computerized scan of the brain was done which had the following findings: dilatation of the left occipital horn of the lateral ventricle, partial agenesis of the corpus callosum, hypoplastic cerebral vermis with lower portion of the fourth ventricle communicating with cisterna magna, widening of the CSF cisterna at left fronto-temporal region. He was noted at day five of life, to have a grade 3/6 ejection systolic murmur at the upper sternal boarder and loud second heart sound. The chest X-ray was unremarkable and the 12 leads electrocardiogram was consistent with right ventricular hypertrophy. Echocardiogram was performed and it showed severe coarctation of the aorta at the level of isthmus and a large patent ductus arteriosus with bi-directional shunt flow mainly right to left. He underwent a successful coarctation repair at day five of life. He is currently

six months old, seizures free on phenytion with developmental delay.

DISCUSSION

Delleman syndrome or oculocerebrocutaneous syndrome^[1], is a rare genetic disorder characterized by eye abnormalities, skin abnormalities, areas of alopecia in combination with hydrocephalus^[2-4]. It is thought that this condition usually occurs sporadically^[5]. In this report we presented the findings in our case and compared it with a previously reported cases (Table 1). In our case

Table 1
Features of Delleman syndrome

Previously reported cases (n = 24)		Our case
Gender (F: M)	8 : 13	M
Developmental delay/MR	14/16	+
Convulsion/EEG abnormalities	12/17	+
Asymmetry	13/14	+
Ocular		
Orbital cyst	18/23	-
AN/microphthalmia	17/21	+
Eyelid coloboma	8/16	+
Hamartoma	6/13	-
Skin		
Periorbital/facial appendages	22/23	+
Hypoplasia	19/20	+
Skeletal		
Skull defects	10/18	+
Rib dysplasia	7/17	-
CT scan		
Intracranial cysts	14/18	+
Agenesis of corpus callosum	9/18	+
Cardiovascular Coarctation of aorta	None	+

M = male; F = female; + = present; - = absent
Table adapted from Hennekam^[6] and Moog *et al*^[7].

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there is an association of a congenital heart disease i.e., coarctation of aorta with Delleman Syndrome; and to our knowledge this is the first time that such an association is reported.

SUMMARY

Any patient with Delleman syndrome should have a detailed echo-cardiography study to rule out an associated congenital heart disease.

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