

13. Munoz A, Leenhardt A, Saisine A, *et al.*
Therapeutic use of adenosine for termi-
nating spontaneous paroxysmal suprav-

tricular tachycardia. *Eur Heart J* 1984, 5:
735-738.

Truncus Arteriosus and Depressor Anguli Oris Muscle Deficiency

P.M.C. Nair
N. Venugopal
Anitha
Razia Begum
N. Sathy

Truncus arteriosus is described along with facial dysmorphism because of the defective development of II and IV branchial arches. This indicates embryological insult between the fourth and seventh weeks of gestation. However, truncus with depressor anguli oris muscle deficiency and polysyndactyly have not been reported. We report such a case in a newborn baby.

Case Report

A one-day-old male baby born to a

*From the Newborn Unit, Department of Pediatrics,
S.A.T. Hospital, Medical College, Thiru-
vananthapuram.*

*Reprint requests: Dr P.M.C. Nair, Associate
Professor (NC) Pediatrics, S.A.T. Hospital,
Medical College, Thiruvananthapuram
695 011.*

*Received for publication: October 13, 1993;
Accepted: November 8, 1993*

nonconsanguinous primigravida mother at 36 weeks of gestation by normal vaginal delivery, was brought to our Neonatal Unit with history of not sucking well. There was no history of any abortion, radiation or drugs in the first trimester of pregnancy. The mother did not suffer from diabetes or any other illness.

The baby weighed 2.2 kg and was tachypneic. There was depressor anguli oris muscle deficiency on the left side (*Fig. 1*) and polydactyly in both the lower limbs. The cardiovascular system examination showed mild cardiomegaly, normal heart sounds and an ejection systolic murmur of Grade III intensity over the left lower sternal border. The liver was palpable 4 cm below the right costal margin in the mid-clavicular line. X-ray chest showed cardiomegaly (CT ratio 62%) and thymic shadow could be appreciated in the X-ray. Serum calcium was normal. The echocardiographic findings were diagnostic of type I truncus, large atrial septal defect and subaortic ventricular septal defect. The baby was treated symptomatically and planned for cardiac surgery at a later date.

Discussion

Persistent truncus arteriosus is an uncommon cardiovascular malformation accounting for 1 to 4% of cardiac deformities found in a number of large autopsy series(1). The defect results from failure of septation of embryonic truncus by the infundibular



Fig. 1. Asymmetric crying facies—Depressor anguli oris muscle deficiency on left side.

truncal ridges. It is always accompanied by a ventricular septal defect. Infants with truncus arteriosus are sometimes said to have a characteristic facies which consists of hypertelorism, downward slanting of palpebral fissures, low set ears, micrognathia a short philtrum and a small mouth(1). Di-George anomaly primarily involves 3rd and 4th pharyngeal arches resulting in associated defects of parathyroid and thymus glands.

Congenital hypoplasia of depressor anguli oris muscle is a well described syndrome in which lower lip on one side fails to be depressed on crying resulting in an "asymmetric crying facies". The normal function of the depressor anguli oris muscle is to draw the lower corner of lip downwards and evert it, so that in its deficiency or hypoplasia, the lower lip on the abnormal side remains unaltered in position while

the baby cries with the angle of mouth being pulled down to the sound side due to unopposed action of depressor anguli oris muscle(2,4).

The importance of identifying this anomaly is that it is associated with other congenital malformations in over 20% of cases, most commonly being associated with cardiovascular anomalies and congenital dislocation of hip. Of the 44 infants with this syndrome, Pape and Pickering found 27 to have major anomaly of skeletal, genitourinary, respiratory and cardiovascular systems. The disorder most commonly associated with this facial defect is congenital heart disease, the commonest defect being ventricular septal defect(2,4).

In this baby, depressor anguli oris muscle deficiency was associated with congenital heart disease in the form of truncus arteriosus and other anomalies such as polydactyly, suggesting an embryonic defect

affecting multiple organ system. To our knowledge the combination of defects which we have described in this baby has not been reported in literature.

REFERENCES

1. Radford D.T. Truncus arteriosus and facial dysmorphism. *Aust Pediatr J*1985,21: 131-133.
2. Menkes HJ. Congenital hypoplasia of the Depressor Anguli Oris muscle. *In: Schaffer and Avery 'Diseases of the Newborn'*. 6th edn. Eds Taeusch HW, Ballard RA, Avery ME. Philadelphia, WB Saunders Co, 1991, p 410.
3. Millen SJ, Baruah JK. Congenital hypoplasia of the depressor anguli oris muscle in the differential diagnosis of facial paralysis. *Laryngoscope*, 1983,93:1168-1170.
4. Singhi S, Singhi P, Lall KB. congenital asymmetrical crying fades. *Clin Pediatr* 1980, 19: 673-675.

Alkaptonuria: Early Detection

R. Khadagawat
R. Teckchandani
P. Garg
A. Arya
B. Choudhary

Alkaptonuria is an inborn error of tyrosine metabolism resulting from deficiency of the enzyme homogentisic acid oxidase which is necessary for converting homogentisic acid into malylaceto-acetic acid(1,3).

Case Report

A one-month-old boy born to non-consanguinous normal parents, was brought with the complaint of slight alteration in urine color. He was the product of full term normal delivery. His weight, length, arm-span and other anthropometric measurements, as well as general and systemic examination revealed no abnormality.

From the Departments of Pediatrics, J.L.N. Medical College, Ajmer.

Reprint requests: Dr. Pukhraj Garg, Jain Colony, Madanganj-Kishangarh 305 801. Ajmer, Rajasthan.

Received for publication: June 21, 1993;

Accepted: November 11, 1993