Clinical presentation of isolated transposition of the great arteries in a complete population based cohort. Implications for pre- and postnatal screening routines.

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Objectives: To determine to which degree pulse oximetry screening (POS) contribute to an earlier diagnosis of isolated transposition of the great arteries (TGA) and to estimate the proportion of neonates with TGA who would benefit from a prenatal diagnosis.

Methods: Infants with TGA born in our referral area for pediatric cardiac surgery from 2003-01-01 to 2013-08-01 were identified from our surgical files and from the causes of death registry (National Board of Health and Welfare). Data on clinical presentation, diagnosis, management and outcome were collected from hospital charts.

Results: 91 cases were identified of which 34 were born in hospitals using POS. 3 were diagnosed prenatally. 57 developed early symptoms and were diagnosed before routine newborn physical examination and before POS. 7 were detected by POS, 13 at the routine newborn physical examination, 1 after that examination but before discharge and 10 were discharged undiagnosed. Five of those discharged were detected at a routine follow-up visit (one at 6 weeks of age) and 5 came to hospital because of symptoms. None of 34 born in hospitals with POS were discharged undiagnosed compared to 10 of 57 in the remaining hospitals. The age at suspicion of congenital heart disease was significantly lower in those born in hospitals with POS. 61 underwent balloon atrial septostomy (BAS), in 9 cases before 6 hours of age and in 26 cases before 12 hours. One neonate died at 4 hours of age at the referring hospital after an unsuccessful BAS. There was no further deaths at a median follow-up of 4.4y (30d-10y). 9 children had neurological symptoms at follow-up of which 3 had neurological symptoms (seizures) already before the arterial switch operation. These three neonates had severe hypoxia (20-50%) and high levels of lactate at arrival at our centre. One needed CPR immediately after arrival. BAS was performed at 6, 8 and 14 hours of age respectively.

Conclusions: Although POS prevents discharge of infants with undiagnosed TGA, at least 4 of 91 (all born in hospitals using POS) would have potentially benefited from a prenatal diagnosis to avoid death or neurological sequelae.