

through the initial steps. This is in contrast to the recommendations of ET suctioning for non-vigorous babies. Even though there is no evidence to support or refute the practice of ET suctioning in non-vigorous babies, the current NRP guidelines do not actually recommend a change in the practice. It will be useful to actually test and validate the above changes in the algorithm in the field for different level of health personnel. Third, assessment based on color has been removed and is replaced by the use of pulse oximetry for the assessment of oxygenation. It is also stated that "oximetry be used when resuscitation can be anticipated, when positive pressure is administered for more than a few breaths, when cyanosis is persistent, or when supplementary oxygen is administered". NRP recommends switching over to 100% oxygen if no improvement occurs in room air after 90s of resuscitation. If pulse oximeter has to be attached in these selective situations, which will be about 30s after birth, it may take up to 90 more seconds for the pulse oximeter signal to appear [2]. By that time the resuscitation will be over in majority of the cases and one will not get a chance to titrate FiO₂ with the blender as per the set SpO₂ limits. Fourth, NRP recommends switching over to 100% oxygen in case the heart rate falls below 60bpm. However, it does not mention about absence of improvement indicated by persistence of heart rate in the 60-100 range even after 90s of resuscitation. It would be prudent to recommend an increase in the oxygen concentration even in the latter situation.

Developing nations contribute to the majority of the neonatal mortality and morbidity due to perinatal asphyxia. Yet, most of the delivery rooms and resuscitation corners in these countries are not equipped with air-oxygen blenders and pulse oximeters [3]. It would be a mammoth, long drawn and expensive task to ensure availability of air-oxygen blenders and motion-resistant low perfusion latest generation pulse oximeters in all delivery areas. There is an urgent need to develop consensus guidelines for our own country keeping in mind the ground realities, and also to produce low cost blenders and pulse oximeters.

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REFERENCES

1. Kattwinkel J, Perlman JM, Aziz K, Colby C, Fairchild K, Gallagher J, *et al.* American Heart Association. Neonatal resuscitation: 2010 American Heart Association guidelines for cardiopulmonary resuscitation and emergency cardiovascular care. *Pediatrics*. 2010;126:e1400-13.
2. O'Donnell CP, Kamlin CO, Davis PG, Morley CJ. Feasibility of and delay in obtaining pulse oximetry during neonatal resuscitation. *J Pediatr*. 2005;147:698-9.
3. Lee AC, Cousens S, Wall SN, Niermeyer S, Darmstadt GL, Carlo WA, *et al.* Neonatal resuscitation and immediate newborn assessment and stimulation for the prevention of neonatal deaths: a systematic review, meta-analysis and Delphi estimation of mortality effect. *BMC Public Health*. 2011;11Suppl 3:S12.

Diagnostic Dilemma in Overlapping Congenital Syndromes

Chromosomal or segmental aneusomy are an important cause of congenital malformations, emphasizing the need for cytogenetic evaluation. Many congenital malformations, especially those with multi-systemic anomalies present overlapping phenotypic features that could partly be attributed to multiple gene deregulations. Moreover, the expressivity of phenotypic features of a particular syndrome could vary extensively among the patients and hence, request for a specific test becomes difficult as observed in the present case.

A 9½-months-old, phenotypically female child was born at term to non-consanguineous parents with a birth

weight of 2700g. She presented with developmental delay and showed microcephaly (<2SD deviation), hypotonia, truncal ataxia, depressed nasal bridge with long philtrum, mild frontal bossing and hepatomegaly of 2.5 cm. Echocardiogram revealed large Ventricular Septal Defect with pulmonary arterial hypertension and a small patent foramen ovale. There was no submucous cleft palate. Developmental assessment suggested a moderate delay with motor development of 4.7 months and mental development of 5.5 months. Other investigations such as TORCH, serum calcium and parathyroid hormone levels were within the normal range. There was no ultrasonographic evidence of renal, urethral and bladder anomaly. Based on these constellations of clinical symptoms and signs, a clinical assessment of 22q11.2 deletion syndrome encompassing DiGeorge syndrome (DGS) was made.

DGS is a common congenital disorder, where pathogenesis has been linked with chromosome 22q11.2

abnormalities [1-3]. Fluorescence *in situ* hybridization (FISH) analysis was carried out using TUPLE region probe (from Kreotech Diagnostics, Netherland) on metaphase and interphase cells. Presence of two intact signals on chromosome 22 ruled out 22q11.2 deletion. Thus, chromosomal analysis was carried out using the GTG-banding technique and the patient was found to be tetrasomy for sex chromosome-X i.e. 48,XXXX.

The degree of clinical presentation for tetrasomy X is highly variable, and tend to have distinctive facial features that include - epicanthal folds, flat nasal bridges, midface hypoplasia, cleft or high arched palates, hypotonia and cardiovascular defects as well as developmental and motor delays [4]. All the above mentioned features can also be observed in cases with 22q deletions as seen in the present study and hence, if only FISH study was processed, tetrasomy X would not have been diagnosed.

This demonstrate that FISH can detect only targeted anomalies whereas conventional cytogenetic can give information about the whole genome alterations and hence be a guide for further diagnostic modalities if required.

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REFERENCES

1. McDonald-McGinn DM, Goldmuntz E, Sullivan K, Eicher P, Gerdes M, Moss E, *et al.* The 22q11.2 Deletion: Screening, Diagnostic Workup, and Outcome of Results; Report on 181 Patient. *Genet Test.* 1997;1:99-108
2. Bartsch O, Nemecková M, Kocárek E, Wagner A, Puchmajerová A, Poppe M, *et al.* DiGeorge/velocardiofacial syndrome: FISH studies of chromosomes 22q11 and 10p14, and clinical reports on the proximal 22q11 deletion. *Amer J Med Genet Part A.* 2003;117:1-5.
3. Tabith Júnior A, Genaro KF, Trindade Júnior AS. Velocardiofacial syndrome with facial and pinna asymmetries. *Braz J Med Biol Res.* 1996;29:1445-7.
4. Schinzel A. Catalogue of Unbalance Chromosome Aberrations in Man. 2nd Edition. New York: Wlatter de Gruyter; 2001.p. 933.

Are Fathers Catching up with Mothers in Liver Donation?

In the last decade, pediatric liver transplantation (LT) has become established therapy for liver failure in our country [1-3]. With growing awareness about the success of LT and safety of the donor operation, more parents are willing to act as donors. It is believed that in India there is apprehension amongst the male members of the family to come forward for kidney donation [4]. To study whether there is any difference in donor demographics pertaining to liver transplantation we carried out a retrospective review.

A total of 46 pediatric living related liver transplants where a parent was the donor were performed between 1998 and May 2012. The mother was the donor in 25 (8 pre-2006, 17 post-2008) cases and the father was the donor in 21 cases (3 pre-2006, 18 post-2008). Post 2006 the proportion of fathers as donors increased from 27.3 % to 51.4%, whereas the proportion of mothers decreased from 72.7% to 48.6%. There was a significant ($P < 0.01$) difference in the sex ratio of the parental donor when compared between the two eras. The year 2007 was chosen as the cut off between two eras because it is considered as a

watershed in our transplant program with a substantial increase in the number of transplants and also it marked a decade of successful liver transplantation in India. Although the donors are decided on the basis of their anatomic suitability to donate, of late there has been an increase in the proportion of fathers as donors. This could be due to greater acceptability of transplantation. The factors responsible for this very welcome development need to be studied.

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REFERENCES

1. Kelly DA, Sibal A. Liver transplantation in children. *Indian Pediatr.* 2006;43:389-91.
2. Poonacha P, Sibal A, Soim AS, Rajashekar MR, Rajakumari DV. India's first successful pediatric liver transplant. *Indian Pediatr.* 2001;38:287-91.
3. Kaur S, Wadhwa N, Sibal A, Jerath N, Sasturkar S. Outcome of live donor liver transplantation in Indian children with bodyweight <7.5 kg. *Indian Pediatr.* 2011;48:51-4.
4. Bal MM, Saikia B. Gender bias in renal transplantation: are women alone donating kidneys in India? *Transplant Proc.* 2007;39:2961-3.