A newborn with partial trisomy 10q and diminutive pulmonary arteries in DORV with critical right ventricular outflow tract obstruction and major aortopulmonary collateral arteries: How invasive to treat?

Kubicki R. (1), Grohmann J. (1), Jakob A. (1), Kroll J. (2), Stuhrmann S. (3), Stiller B. (1)
Department of Congenital Heart Disease, University Heart Centre Freiburg, Germany (1), Department of Cardiovascular Surgery, University Heart Centre Freiburg, Germany (2), Childrens Hospital, Karlsruhe, Germany (3)

Background:
Distal 10q trisomy is an extremely rare chromosomal disorder usually caused by an unbalanced translocation, including a distinctive, dysmorphic appearance, growth retardation and psychomotoric disorders. Patients with trisomy 10q24->qter tend to be associated with cardiac, renal and ocular abnormalities. However, the severity varies greatly among affected individuals. Ethical aspects should therefore be carefully considered, but early rehabilitation is recommended in the setting of diminutive pulmonary arteries (PAs) to achieve favourable PAs growth and the goal of biventricular repair in the future.

Case and results:
Mother 1G1P, no consanguinity, 40 SSW, birth weight 2765 g. The female newborn presented multiple dysmorphic craniofacial features including microstomia, a receding chin with a laryngeal aberrance, dysplastic, low-set ears, an epicanthic fold, and blepharophimosis along with microphthalmia. She also had musculoskeletal manifestations such as muscular hypotonia, talipes calcaneus and contracted joints affecting hands and limbs. Chromosomal analysis showed an unbalanced translocation of 10q [(t1;10), 10q25.2->10qter].
Echocardiography revealed a double outlet right ventricle (DORV) with critical right ventricular outflow tract obstruction (RVOTO) and extremely diminutive pulmonary arteries (PAs) with competitive major aortopulmonary collateral arteries (MAPCAs). To open the right ventricular outflow tract (RVOT) and subatretic pulmonary valve, we decided on initial palliation with balloon dilatation on day 7, followed by RVOT stent implantation eight weeks later. Repeated catheterisation with stent dilatation, valvuloplasty and embolism in the MAPCAs showed adequate PAs growth.
The child developed satisfactorily and underwent specific early ophthalmic and orthopaedic treatments. Parental bonding developed very well. Biventricular repair with a 12 mm Contegra valve was successful at the age of 14 months in the presence of well-developed PAs.

Conclusion:
Instead of early high-risk unifocalisation, this early interventional approach with RVOT stent and repeated interventional cardiac catheterisation was a safe and effective treatment strategy that encouraged PAs growth. This approach provided time for early therapy of extracardiac dysmorphisms and for the family’s ethical considerations.