Metric #: 020
Effective: 01.10.2017

Genetic Testing in Tetralogy of Fallot Patients	
Measure Description: Proportion of Tetralogy of Fallot (ToF) patients who received a test for 22q11.2 deletion.	
Numerator	Number of ToF patients who received or had an order for 22q11.2 deletion testing any time in their medical history.
Denominator	Number of patients, ≤ 18 years old, with ToF who had a visit during the measurement period.
Denominator Exclusions	• Patient or parent refusal • Patients with repaired TOF with A-V canal, Pulmonary Atresia/MAPCAS or TOF with absent valve. • Other known genetic diagnoses (e.g Trisomy 21, 13, 18 and Alagille syndrome)
Denominator Exceptions	None
Definitions/Notes	None
Measurement Period	Quarterly
Sources of Data	Medical Record
Attribution	This measure should be reported by qualified providers with experience and expertise in this modality
Care Setting	Outpatient

Rationale

These measures are meant to be applied to all patients with a 'typical' tetralogy of Fallot repair and may not be suitable for those smaller groups with more complex subtypes. Repaired TOF patients with A-V canal, Pulmonary Atresia/MAPCAS or TOF with absent valve will be excluded. Those with major underlying genetic disorders (e.g. Trisomy 21, 13, 18;) will also be excluded from this set of measures.

Patients with TOF can have significant associated genetic syndromes or chromosomal anomalies in up to 25% of cases, including trisomies 21, 18 and 13, Alagille syndrome and others. Up to 15% of cases of ToF have 22q ll.2 deletion (including 6% in those with normal aortic arch and branching). This testing is important as it can have implications on the management of the patient as well as on the counseling of the family.

Clinical Recommendation(s)

ACC/AHA Guidelines

1. Wamcs CA, Williams RG, Bashore TM, Child JS, Connolly HM, Dearani JA, del Nido P, Fasulcs JW, Graham TP, J r., l lijazi ZM, Hunt SA, King ME, Landzberg MJ, Miner PD, Radford MJ, Walsh EP, Webb GO, Smith SC, Jr., Jacobs AK, Adams CD, Anderson JL, Antman EM, Buller CE, Creager MA, Ettinger SM, Halperin JL, Krumholz liM, Kushner FG, Lytle BW, Nishimura RA, Page RL, Riegel B, Tarkington LG, Yancy CW. Ace/aha 2008 guidelines for the management of adults with congenital heart disease: A report of the American College of Cardiology/American Heart Association task force on practice guidelines (writing committee to develop guidelines on the management of adults with congenital heart disease). Developed in collaboration with

the American Society of Echocardiography, Heart Rhythm Society, International Society for Adult Congenital Heart Disease, Society for Cardiovascular Angiography and Interventions, and Society of Thoracic Surgeons. Journal of the American College of Cardiology. 2008;52:c1 43-263

Other guidelines:

1. Pierpont MEet al. Genetics of congenital Heart defects: current knowledge: a scientific statement from the American Heart Association, council on Cardiovascular Disease in the Young. Circulation 2007; 11 5:3015-38.
2. Silversidcs CK, Kiess M, Beauchesne L, Bradley T, Connell y M, Niwa K, Mulder B, Therrien J. Canadian Cardiovascular Society 2009 Consensus Conference on the management of adults with congenital heart disease: outflow tract obstruction, coarctation of the aorta, tetralogy of Fallot, Ebstein anomaly and Marfan'ssyndrome. Qill.1 2010 Mar;26(3):e80-97.
3. Mom ma K, Takao A, Matsuoka R, et al. Tetralogy of Fallot associated with chromosome 22q11 .2 deletion in adolescents and young adults. Genet Mcd. 2001; 3:56-60.
4. Fahed ACet al. Genetics of congenital heart disease: the glass half empty. Circ Res 201 3; 11 2:707-20.
5. Amati F, MariA, Digilio MC, Mingarelli R, Marino 13, Giannotti A, Novelli G, Dallapiccola B. 22q l l deletions in isolated and syndromic patients with tetralogy of Fallot. /hun Genet. 1995; 95:479-482.
6. Goldmuntz E, Clark BJ, Mitchel LE et al. Frequency of22g II deletions in patients with conotruncal defects. J ACC 1998; 32:492-498

Challenges to Implementation

Data collection, submission and database management costs

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