ACPC Quality Network

22q11 Testing in Tetralogy of Fallot

ACPC QNet

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The Metric: 22q11.2 Testing in Tetralogy of Fallot

- Number of patients ≤ 18 yoa with Tetralogy of Fallot (TOF), who have undergone/ been ordered for 22q11.2 deletion testing
- Exclusions:
  - More complex forms of TOF
    - TOF/AV canal
    - TOF/pulmonary atresia with MAPCAS
    - TOF/absent pulmonary valve
  - Other genetic disorders
    - Trisomy 21, 13, 18
    - Alagille syndrome

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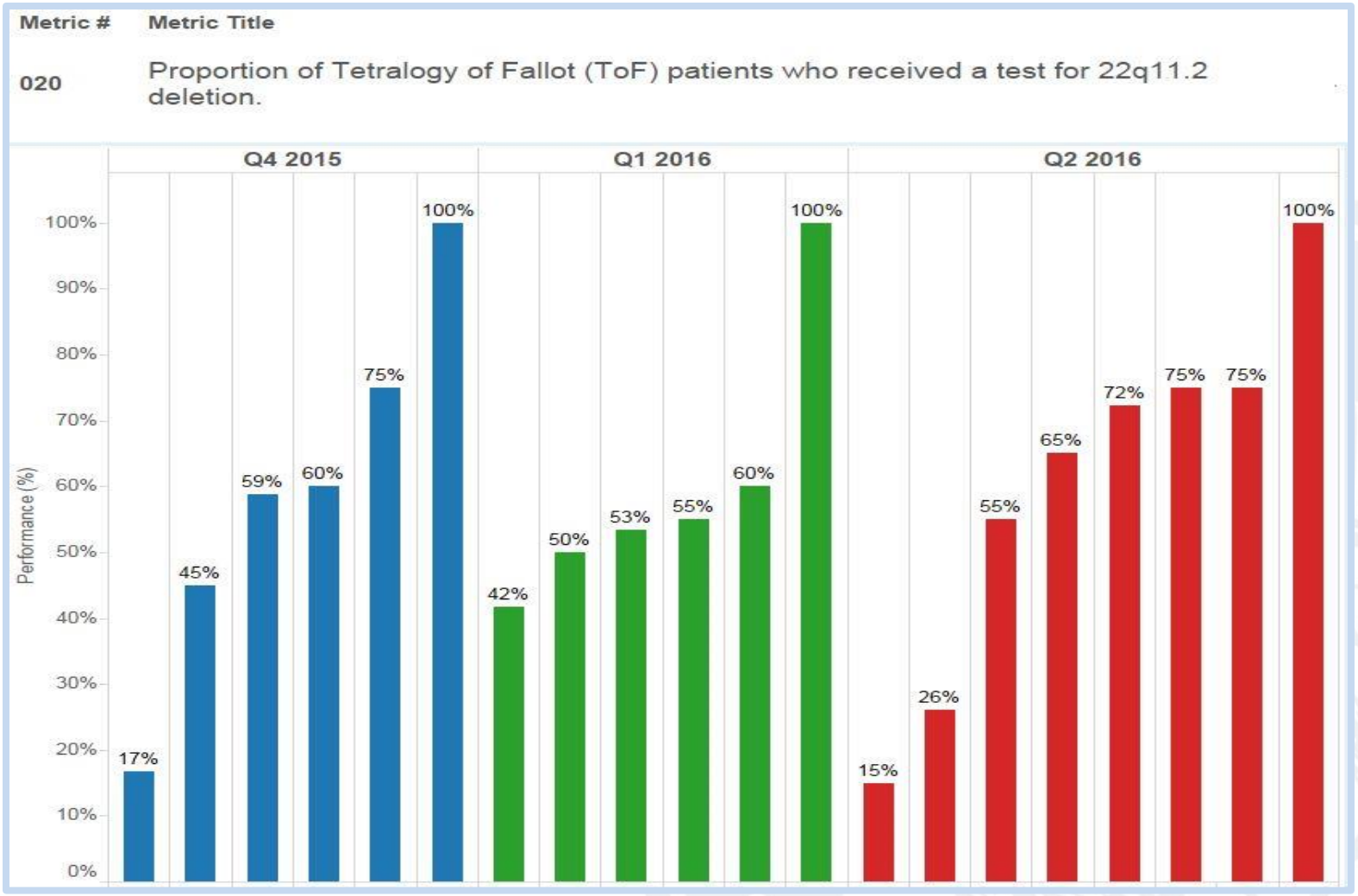
Why Test for 22q11.2 in TOF?

- TOF patients can have significant genetic findings in up to *25% of cases*
  - Trisomy 21,13, or 18, and Alagille syndrome
  - 22q11.2 deletion present in:
    - 15% of all TOF patients
    - 6% with left aortic arch with normal branching
- Knowledge of 22q11.2 deletion can impact:
  - Clinical management
  - Counseling

Why Test for 22q11.2 in TOF?

- Impact on care with knowledge of 22q11:
  - Neurocognitive
  - Feeding difficulties
    - Palate
    - Motility
  - Immune deficiency
  - Endocrine:
    - Parathyroid - calcium regulation
    - Thyroid
  - Thrombocytopenia
  - Renal anomalies
  - Psychiatric disease
  - Genetic counseling – autosomal dominant

What has the data shown so far?



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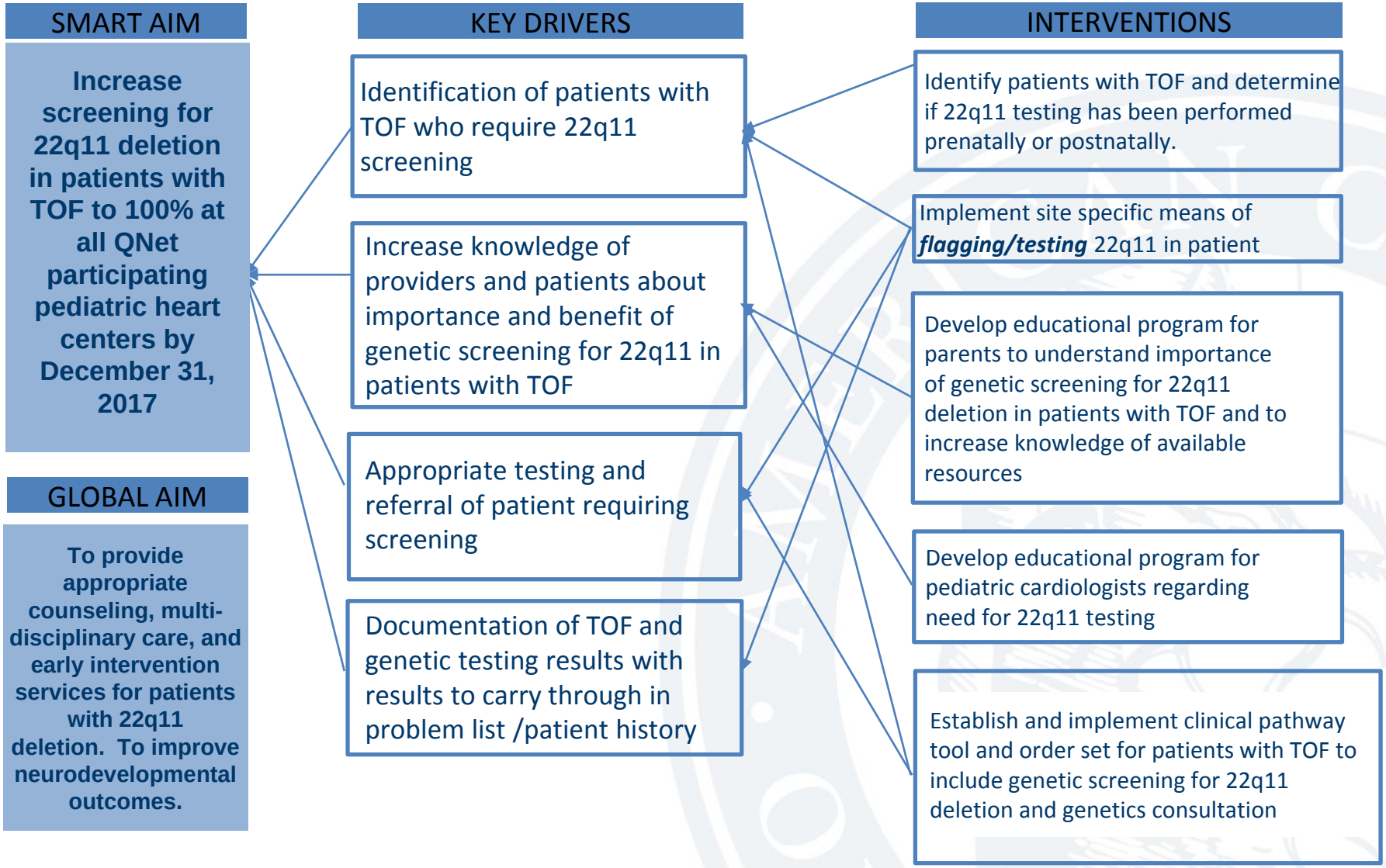
Why is Testing for 22q11.2 so Variable?

- Practice variation
  - Only ordered if suspected versus in all
  - Cardiology versus genetics
- Difficulty in data capture
  - Genetic testing sent prenatally
  - Genetic testing sent from hospital of birth
- Variable documentation of data
  - Consistent location in medical record
  - Record even if normal

22q11 Screening in TOF: KEY DRIVER DIAGRAM



22q11 Screening in TOF: KEY DRIVER DIAGRAM

SMART AIM

Increase screening for 22q11 deletion in patients with TOF to 100% at all QNet participating pediatric heart centers by December 31, 2017

GLOBAL AIM

To provide appropriate counseling, multi-disciplinary care, and early intervention services for patients with 22q11 deletion. To improve neurodevelopmental outcomes.

KEY DRIVERS

Identification of patients with TOF who require 22q11 screening

INTERVENTIONS

Identify patients with TOF and determine if 22q11 testing has been performed prenatally or postnatally.

Implement site specific means of ***flagging/testing*** 22q11 in patient

Establish and implement clinical pathway tool and order set for patients with TOF to include genetic screening for 22q11 deletion and genetics consultation

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KEY DRIVERS

Increase knowledge of providers and patients about importance and benefit of genetic screening for 22q11 in patients with TOF

INTERVENTIONS

Develop educational program for parents to understand importance of genetic screening for 22q11 deletion in patients with TOF and to increase knowledge of available resources

Develop educational program for pediatric cardiologists regarding need for 22q11 testing

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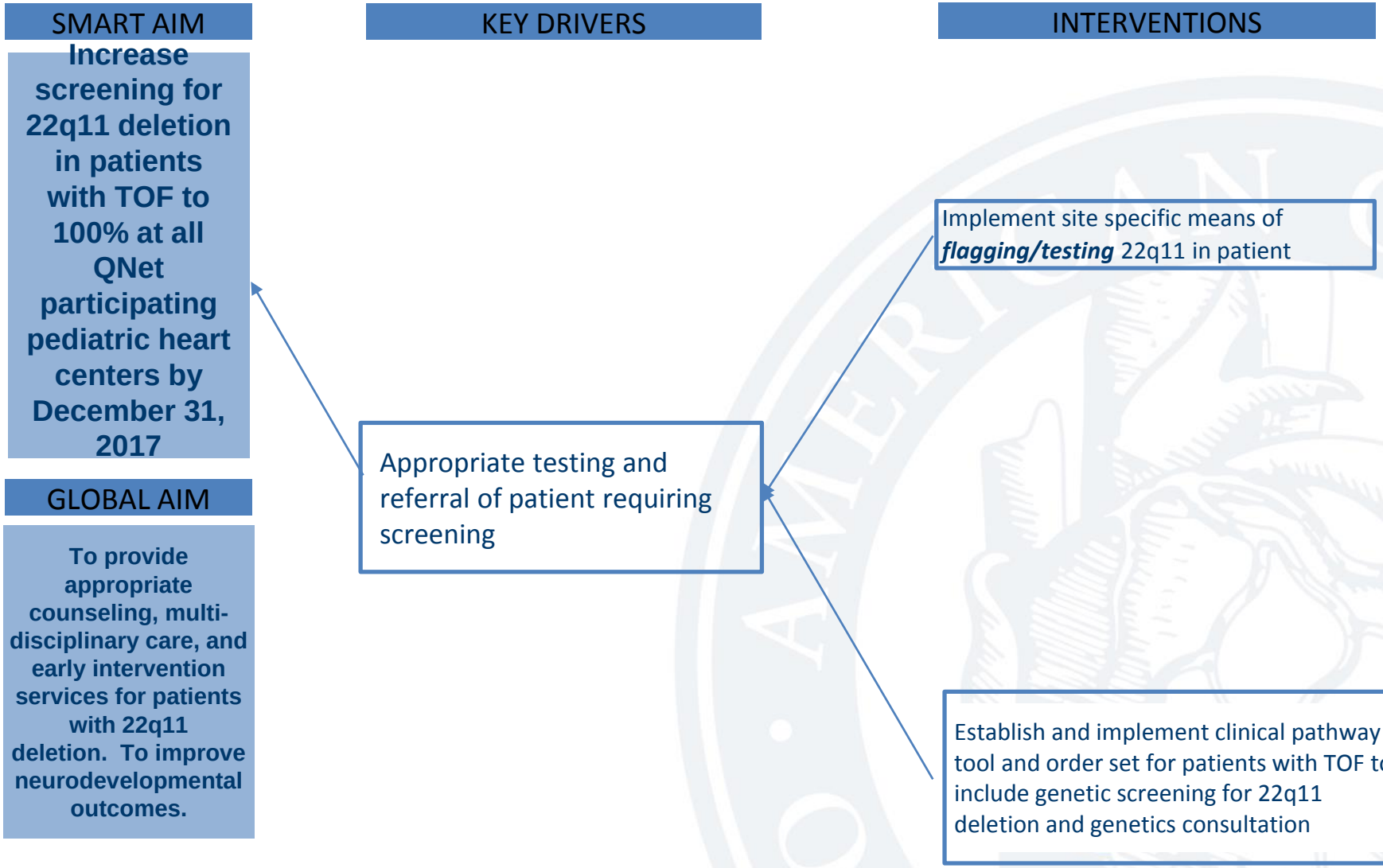

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KEY DRIVERS

Documentation of TOF and genetic testing results with results to carry through in problem list/patient history

INTERVENTIONS

Implement site specific means of ***flagging/testing*** 22q11 in patient

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Next Steps

- Early implementation of strategies at various centers
  - Assess successes
  - Assess hang-ups
- Talk to institution “F”!!!
- Re-strategize interventions
- Track changes in quarterly data

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Thank You!

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