QNet Quality Improvement:
22q11 Deletion Syndrome testing in Tetralogy of Fallot

22q11 Deletion Syndrome (aka DiGeorge syndrome, Velocardiofacial syndrome, CATCH 22)

- Approximately 15% of individuals with TOF have 22q11 DS
- 22q11DS can cause extra-cardiac and developmental complications but is very variable. Though there is a long list of potential co-morbidities, no individual has all of them and there is a spectrum from mild to severe for each potential complication.
- Potential extra-cardiac complications:
  - Feeding difficulties
  - Immune deficiency
  - Growth delay
  - Cleft palate
  - Hypocalcemia
  - Delayed development
  - Learning, behavioral, and psychiatric issues

Reasons to test:
- Allow for initial screening and long term monitoring for potential complications
- Improve neurodevelopmental outcomes by offering early, individualized developmental services
- Provide accurate recurrence risk information for families
- Allow referral for ongoing support and information resources

How to test:
- In PowerChart “22q11.2/10p13-14 Analysis DNA”
  - Clinical Information: Congenital Heart Disease
  - Diagnostic/Non Diagnostic: Diagnostic
  - Specimen Type: Blood
- Will the test be covered? This is clinically indicated, diagnostic genetic testing. However, families should be reminded that they are still responsible for their annual deductible (if not yet met) and co-pay
- Can my child be denied insurance if the test is positive? The Genetic Information Nondiscrimination Act (GINA) is federal legislation that makes it illegal for an insurance company to deny coverage based on the diagnosis of a genetic disorder. There are, however, no such protections for life insurance.

If the test has been done and is positive:
Refer to Cardiovascular Genetics for a detailed discussion of the diagnosis and for longitudinal follow up and care