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