SB1612 was passed in 2009 to increase awareness of Velo-Cardio-Facial (a.k.a. DiGeorge or 22q11.2 deletion) syndrome in Texas. As a result parents whose children are receiving services for developmental delays through the state's Early Childhood Intervention (ECI) programs may be contacting their pediatricians to ask whether their child may need genetic testing for the 22q11.2 deletion.

DOES YOUR PATIENT HAVE VCFS???

Velocardiofacial syndrome or DiGeorge syndrome (VCFS) is a common condition which occurs when there is a small deletion of chromosome 22 (22q11.2 deletion). It is the second most common genetic disorder after Down syndrome, occurring in 1 in 2,000 births.

Signs and symptoms may include:

- Long face
- Feeding difficulties
- Speech and other developmental delays
- Heart defect
- Infections
- Tapered digits

For more information, visit:
www.chop.edu/consumer/jsp/division/generic.jsp?id=74637.

Diagnose the deletion by a simple blood test called a chromosome microarray analysis. This test can detect deletions and duplications of chromosome 22 among others which can also present with similar signs and symptoms. Information on this test and one of the labs offering this service can be found at:
www.bcm.edu/geneticlabs/tests/cyto/CMA.html.

Treatment for VCFS is targeted to the symptoms of the individual patient and should start early to maximize clinical outcomes. If the diagnosis is suspected, or once a diagnosis of VCFS is confirmed by laboratory analysis, the patient should be referred to a medical geneticist for evaluation, counseling, and management.

Educational brochure about VCFS: www.vcfsef.org.
Texas support group: www.vcfstexas.com.