Diagnostic Criteria

Patient has…

- ◯ DiGeorge defined genetically ONLY
- ◯ Classic clinical triad for complete DiGeorge syndrome
- ◯ Combination of genetic and phenotypic features

Defined Genetically Only

Select all that apply

- ☐ Chromosome 22q11.2 deletion
- ☐ TBX mutation
- ☐ Chromosome 10p13-14 deletion
- ☐ CHD7 mutation

Classical Clinical Triad

Select all that apply

- ☐ Conotruncal cardiac anomaly
- ☐ Hypocalcemia
- ☐ T cells < 500 /mm3 in the first 3 months of life

Combination of Features

Select all that apply

- ☐ Chromosome 22q11.2 deletion
- ☐ Conotruncal cardiac anomaly or cardiothoracic vascular anomaly
- ☐ Hypocalcemia
- ☐ Diminished T cell counts for age
- ☐ Hypoplastic thymus visualized
- ☐ Dysmorphic facies
- ☐ Tracheoesophageal fistula
- ☐ Coloboma Cleft palate (frank clefting or submucous cleft)
- ☐ Velopharyngeal insufficiency / hypernasal speech

Other Questions

Fetal toxin exposure?

- ◯ Yes
- ◯ No
- ◯ Unknown

(if yes) Explain:

- ☐ Maternal diabetes
- ☐ Alcohol exposure
- ☐ Isotretinoin exposure
- ☐ Other: _____