



**DiGeorge Syndrome (DGS) Registry Data Collection Form**

**Patient Identification:**

Patient Name (first, middle, last) \_\_\_\_\_

Date of this Record (mm/dd/yyyy): \_\_\_\_/\_\_\_\_/\_\_\_\_.

Patient's Number Assigned by Registry \_\_\_\_\_

Is this the initial registration of this patient[  ] or follow-up?[  ]

Patient Initials \_\_\_\_/\_\_\_\_/\_\_\_\_ Date of Birth \_\_\_\_/\_\_\_\_/\_\_\_\_ Gender: male [  ], female [  ]

Home Address: \_\_\_\_\_

State: \_\_\_\_\_

Zip Code: \_\_\_\_\_

Phone: \_\_\_\_\_

Email: \_\_\_\_\_

State or Province of birth: \_\_\_\_\_

Country of birth: \_\_\_\_\_

Data transferred from earlier IDF Registry? Yes [  ] no [  ]

**Submitting Physician Information:**

Name: \_\_\_\_\_

Address: \_\_\_\_\_

State: \_\_\_\_\_

Zip Code: \_\_\_\_\_

Phone: \_\_\_\_\_

Email: \_\_\_\_\_

Fax: \_\_\_\_\_

**Diagnostic Criteria** please indicate which situation applies

- Defined genetically only (chromosome 22q11.2 deletion, TBX mutation, chromosome 10p13-14 deletion)
- Classic clinical triad for complete DiGeorge syndrome: (Conotruncal cardiac anomaly, hypocalcemia, CD3 T cells < 500 /mm3 in the first 3 months of life)
- Combination of genetic and phenotypic features (Mark 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

**DGA Gene Mutation**

(Number nucleotides using Human Mutation 11:1-3, 1998)

Mutation analysis performed by \_\_\_\_\_

- Hemizygous deletion of chromosome 22q11.2
- TBX point mutation (please specify) \_\_\_\_\_
- Hemizygous deletion of 10p13-14
- CHD7 mutation (please specify) \_\_\_\_\_
- Fetal toxin exposure (please specify) (often maternal diabetes, alcohol, isotretinoin) \_\_\_\_\_
- Other (please specify) \_\_\_\_\_

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**Genetic Information**

Sporadic [ ] (no family history) or \_\_\_\_\_ [ ] pattern of inheritance  
 Family history unknown [ ]

**Family history**

Relation	Normal	Not tested	Carrier	DGA – alive	DGA – deceased	Unknown	Undiagnosed with suggestive symptoms
Mother							
Father							
Brothers (fill in # in each category)							
Sisters (fill in # in each category)							
Uncles (fill in # in each category)							
Aunts (fill in # in each category)							
Grandmothers (fill in # in each category)							
Grandfathers (fill in # in each category)							
Cousins (fill in # in each category)							
Other affected family members							

Information on Other Affected Kindred Members listed above

Relationship	Initials	Gender	Date of Birth	Registered in database?	
				Yes	No

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**Other treatments used**

	Yes	No	Age begun	Date begun (mm/dd/yyyy)	Duration	Still needed?	
						Yes	No
Chronic oxygen							
Parenteral nutrition							
Calcium supplements							
Fetal thymus							
Cultured thymic epithelium							

Other? Please explain \_\_\_\_\_

**Surgery (Check all that apply)**

<input type="checkbox"/>	Cardiac repair
<input type="checkbox"/>	Cardiac repair (multiple)
<input type="checkbox"/>	G-tube
<input type="checkbox"/>	Malrotation repair
<input type="checkbox"/>	Palatal repair
<input type="checkbox"/>	T-E fistula repair
<input type="checkbox"/>	Pharyngoplasty
<input type="checkbox"/>	Fundoplication
<input type="checkbox"/>	Other (specify)

**Developmental interventions (Check all that apply)**

<input type="checkbox"/>	Sign language
<input type="checkbox"/>	Speech therapy
<input type="checkbox"/>	Physical therapy
<input type="checkbox"/>	Occupational therapy
<input type="checkbox"/>	Psychiatry/Psychology
<input type="checkbox"/>	Special Education / tutoring
<input type="checkbox"/>	Other (specify)

**Outcome**

**What is the biggest issue this patient faces?** \_\_\_\_\_

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**Additional Clinical Features of Special Interest**

**Check all that apply**

<b><u>Problems in infancy</u></b>
Feeding problems
Nasal regurgitation
Gastroesophageal reflux
Constipation
<b><u>Cardiac disease</u></b>
Interrupted aortic arch type B
Tetralogy of Fallot
VSD
Truncus arteriosus
Other cardiac defect (specify)
Thymus visualized at time of repair? Yes <input type="checkbox"/> No <input type="checkbox"/>
Thymus Normal Size <input type="checkbox"/> Hypoplastic <input type="checkbox"/> Absent <input type="checkbox"/>
<b><u>Endocrine</u></b>
Hypocalcemia requiring more than 2 months of oral supplementation
Transient hypocalcemia
Hypothyroidism
Growth hormone deficiency
<b><u>School</u></b>
Receiving passing grades in regular classes at grade level
Barely passing regular classes at grade level
Receives some special attention
Receives exclusively special education
Institutionalized
For adults: highest grade completed: _____ Performance at most recent level?
<b><u>Neuropsychiatric</u></b>
Anxiety disorder
Attention deficit hyperactivity disorder
Autism/autistic
Bipolar disorder
Cerebellar ataxia
Depression
Obsessive compulsive disorder
Phobias
Schizophrenia
Seizures (not hypocalcemic)
<b><u>Social/behavior</u></b> (for patients over 16 years of age)
Currently holding a job
Performs household chores independently
Drives a car
Dating/married
Smoking
Excessive alcohol use
Illicit drug use

**Additional comments:**

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