



DiGeorge Syndrome (DGS) Registry Data Collection Form

Patient Identification:

Patient Name (first, middle, last) _____

Patient's USIDNET Registry Number assigned after online enrollment _____

Date of Birth ____/____/____ (mm/dd/yyyy) or Year of Birth _____

Gender: male [], female []

Home Address:

Address: _____

State: _____

Zip Code: _____

Phone: _____

Email: _____

State or Province of birth: _____

Country of birth: _____

Date of this Record Completion (mm/dd/yyyy): ____/____/____

Date of Visit (mm/dd/yyyy): __/____/____

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

Submitting Physician Information:

Name: _____

Address: _____

State: _____

Zip Code: _____

Phone: _____

Email: _____

Fax: _____



DiGeorge Syndrome (DGS) Registry Data Collection Form

Diagnostic Criteria

- 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) _____

DiGeorge Syndrome (DGS) Registry Data Collection Form

Patient Initials: ___/___/___

Genetic Information

Sporadic [] (no prior family history) or _____ [] pattern of inheritance
 Family history unknown []

Family history

Please list additional relationships. If more space is needed, please use the Memo section at the end of this form.

Relation	DGS alive	DGS deceased	Normal	Not tested	Carrier	Unknown	Undiagnosed with suggestive symptoms
Mother							
Father							

Information on Other Affected Kindred Members listed above

Relationship	Initials	Gender	Year of Birth	Listed in Registry? Yes/No/Unknown

DiGeorge Syndrome (DGS) Registry Data Collection Form

Patient Initials: ___/___/___

Other treatments used

	Yes	No	Unknown	Age begun		OR Date begun (mm/dd/yyyy)	Duration	Still needed?	
				Mos.	Yrs.			Yes	No
Chronic oxygen									
Parenteral nutrition									
Calcium supplements									
Fetal thymus transplant									
Cultured thymic epithelium transplant									
Other									

Other? Please explain _____

Surgery (Check all that apply)

	Yes	No	Unknown
Cardiac repair			
Cardiac repair (multiple)			
G-tube			
Malrotation repair			
Palatal repair			
T-E fistula repair			
Pharyngoplasty			
Fundoplication			
Other (specify)			

Developmental interventions (Check all that apply)

	Yes	No	Unknown
Sign language			
Speech therapy			
Physical therapy			
Occupational therapy			
Psychiatry/Psychology			
Special Education / tutoring			
Other (specify)			

Outcome

What is the biggest issue this patient faces? _____

DiGeorge Syndrome (DGS) Registry Data Collection Form

Patient Initials: ___/___/___

Additional Clinical Features of Special Interest

Check all that apply

Yes	No	Unknown	<u>Problems in infancy</u>
			Feeding problems
			Nasal regurgitation
			Gastroesophageal reflux
			Constipation
			<u>Cardiac disease</u>
			Interrupted aortic arch type B
			Tetralogy of Fallot
			VSD
			Truncus arteriosus
			Other cardiac defect (specify)
			Thymus visualized at time of repair?
			Thymus Normal Size <input type="checkbox"/> Hypoplastic <input type="checkbox"/> Absent <input type="checkbox"/>
			<u>Endocrine</u>
			Hypocalcemia requiring more than 2 months of oral supplementation
			Transient hypocalcemia
			Hypothyroidism
			Growth hormone deficiency
			<u>School</u>
			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? Satisfactory / Unsatisfactory
			<u>Neuropsychiatric</u>
			Anxiety disorder
			Attention deficit hyperactivity disorder
			Autism/autistic
			Bipolar disorder
			Cerebellar ataxia
			Depression
			Obsessive compulsive disorder
			Phobias
			Schizophrenia
			Seizures (not hypocalcemic)
			<u>Social/behavior</u> (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:
