Appendix B: List of Rare Diseases

Rare caregivers were asked to indicate what rare disease, illness, or condition their care recipient had from a list of over 6,000 potential rare diseases. Below is a table showing the percentage and count of rare caregivers represented in this study by each condition they selected.

	All Rare Caregivers (n=1,406)	
	Percentage	Number
Cystic fibrosis	**9.5%**	133
Pulmonary arterial hypertension	**3.6%**	51
Atypical hemolytic uremic syndrome	**2.1%**	29
Ehlers-Danlos syndromes	**1.8%**	26
Fabry disease	**1.8%**	26
Congenital central hypoventilation syndrome	**1.7%**	24
PANDAS	**1.6%**	23
SATB2-associated syndrome	**1.4%**	20
Myasthenia gravis	**1.4%**	19
Osteogenesis imperfecta	**1.2%**	17
Eosinophilic enteropathy	**1.1%**	16
Hypermobile Ehlers-Danlos syndrome	**1.1%**	16
Hypophosphatasia	**1.1%**	16
Cardiofaciocutaneous syndrome	**1.1%**	15
Huntington disease	**1.1%**	15
Tuberous sclerosis	**1.1%**	15
Prader-Willi syndrome	**0.9%**	12
Hemophilia A	**0.8%**	11
Lennox-Gastaut syndrome	**0.8%**	11
Alternating hemiplegia of childhood	**0.7%**	10
CDKL5-related disorder	**0.7%**	10
Neuronal ceroid lipofuscinosis	**0.7%**	10
Shwachman-Diamond syndrome	**0.7%**	10
Zellweger syndrome	**0.7%**	10
Alpha-1 antitrypsin deficiency	**0.6%**	9
DDX3X-related intellectual disability	**0.6%**	9
Pediatric acute-onset neuropsychiatric syndrome	**0.6%**	9
CADASIL	**0.6%**	8
Primary sclerosing cholangitis	**0.6%**	8
Sickle cell anemia	**0.6%**	8

	All Rare Caregivers (n=1,406)	
	Percentage	Number
Spinal muscular atrophy	0.6%	8
Loeys-Dietz syndrome	0.5%	7
X-linked myotubular myopathy	0.5%	7
Autosomal dominant centronuclear myopathy	0.4%	6
CLOVES syndrome	0.4%	6
Duchenne muscular dystrophy	0.4%	6
Hypoparathyroidism	0.4%	6
Imperforate anus	0.4%	6
Peroxisome biogenesis disorder-Zellweger syndrome	0.4%	6
Rett syndrome	0.4%	6
Acquired hemophilia	0.4%	5
Aplastic anemia	0.4%	5
Cerebrospinal fluid leak	0.4%	5
CHARGE syndrome	0.4%	5
Familial lipoprotein lipase deficiency	0.4%	5
Mucopolysaccharidosis type II	0.4%	5
Necrotizing enterocolitis	0.4%	5
Niemann-Pick disease type B	0.4%	5
Panhypopituitarism X-linked	0.4%	5
Pediatric multiple sclerosis	0.4%	5
Sturge-Weber syndrome	0.4%	5
Acromegaly	0.3%	4
ADCY5-related dyskinesia	0.3%	4
Behçet disease	0.3%	4
Cerebrotendinous xanthomatosis	0.3%	4
Childhood hypophosphatasia	0.3%	4
Common variable immunodeficiency	0.3%	4
Costello syndrome	0.3%	4
Dyskeratosis congenita	0.3%	4
Familial dysautonomia	0.3%	4
GATAD2B-associated neurodevelopmental disorder	0.3%	4
Hemophilia B	0.3%	4
Idiopathic thrombocytopenic purpura	0.3%	4
Mastocytosis	0.3%	4
Mitochondrial disease with severe hypotonia, lactic	0.3%	4
Mitochondrial genetic disorders	0.3%	4
Moebius syndrome	0.3%	4
Noonan syndrome	0.3%	4
Osteogenesis imperfecta type I	0.3%	4

	All Rare Caregivers (n=1,406)	
	Percentage	Number
Osteogenesis imperfecta type III	0.3%	4
Osteogenesis imperfecta type IV	0.3%	4
Phenylketonuria	0.3%	4
Spina bifida	0.3%	4
West syndrome	0.3%	4
X-linked hypophosphatemia	0.3%	4
X-linked periventricular heterotopia	0.3%	4
22q11.2 deletion syndrome	0.2%	3
Alagille syndrome	0.2%	3
Arachnoiditis	0.2%	3
Arthrogryposis multiplex congenita	0.2%	3
Autoimmune polyglandular syndrome type 1	0.2%	3
Cerebral palsy	0.2%	3
Cerebro-costo-mandibular syndrome	0.2%	3
Chronic inflammatory demyelinating polyneuropathy	0.2%	3
Cohen syndrome	0.2%	3
Complex regional pain syndrome	0.2%	3
Congenital adrenal hyperplasia	0.2%	3
Cystinosis	0.2%	3
EEC syndrome	0.2%	3
Evans syndrome	0.2%	3
Glycogen storage disease type 2	0.2%	3
Hemophagocytic lymphohistiocytosis	0.2%	3
Hereditary sensory and autonomic neuropathy type 1E	0.2%	3
Hirschsprung's disease	0.2%	3
Hypomyelination with atrophy of basal ganglia and	0.2%	3
Idiopathic pulmonary fibrosis	0.2%	3
Isovaleric acidemia	0.2%	3
Leigh syndrome	0.2%	3
Mosaic trisomy 9	0.2%	3
Moyamoya disease	0.2%	3
Multiple myeloma	0.2%	3
Narcolepsy	0.2%	3
Peroxisomal biogenesis disorders	0.2%	3
Primary ciliary dyskinesia	0.2%	3
Propionic acidemia	0.2%	3
PTEN hamartoma tumor syndrome	0.2%	3
Ring chromosome 20	0.2%	3
Severe combined immunodeficiency	0.2%	3

	All Rare Caregivers (n=1,406)	
	Percentage	Number
Spinal muscular atrophy type 2	0.2%	3
Trisomy 18	0.2%	3
VACTERL association	0.2%	3
WAGR syndrome	0.2%	3
15q13.3 microdeletion syndrome	0.1%	2
16p11.2 deletion syndrome	0.1%	2
1q21.1 microdeletion syndrome	0.1%	2
Acquired hemophilia A	0.1%	2
Aicardi-Goutieres syndrome	0.1%	2
AL amyloidosis	0.1%	2
Ataxia telangiectasia	0.1%	2
Autoimmune encephalitis	0.1%	2
Bohring-Opitz syndrome	0.1%	2
Carcinoid tumor	0.1%	2
Cerebellar ataxia, areflexia, pes cavus, optic atrophy and	0.1%	2
Chondrodysplasia punctata 1, X-linked recessive	0.1%	2
Chromosome 18q duplication	0.1%	2
Chromosome 4q deletion	0.1%	2
Cornelia de Lange syndrome	0.1%	2
Corticobasal degeneration	0.1%	2
Crohn's disease of the esophagus	0.1%	2
D-bifunctional protein deficiency	0.1%	2
Dementia, familial Danish	0.1%	2
Focal dermal hypoplasia	0.1%	2
FOXG1 syndrome	0.1%	2
Friedreich ataxia	0.1%	2
Frontotemporal dementia	0.1%	2
Gastroschisis	0.1%	2
Gaucher disease	0.1%	2
GM1 gangliosidosis	0.1%	2
Guillain-Barre syndrome	0.1%	2
Hemimegalencephaly	0.1%	2
Hemophilia	0.1%	2
Hurler syndrome	0.1%	2
Hypoplastic left heart syndrome	0.1%	2
Immune thrombocytopenia	0.1%	2
KCNQ2-Related Disorders	0.1%	2
Klippel-Trenaunay syndrome	0.1%	2
Leber hereditary optic neuropathy	0.1%	2

	All Rare Caregivers (n=1,406)	
	Percentage	Number
Leukodystrophy	0.1%	2
Lupus erythematosus tumidus	0.1%	2
McCune-Albright syndrome	0.1%	2
Merkel cell carcinoma	0.1%	2
Metachromatic leukodystrophy	0.1%	2
Mollaret meningitis	0.1%	2
Mucolipidosis type 4	0.1%	2
Mucopolysaccharidosis type I	0.1%	2
Mucopolysaccharidosis type IIIA	0.1%	2
Mucopolysaccharidosis type VI	0.1%	2
Myelodysplastic syndromes	0.1%	2
Neurodegeneration with brain iron accumulation	0.1%	2
Optic nerve hypoplasia, familial bilateral	0.1%	2
Paralysis agitans, juvenile, of Hunt	0.1%	2
Periventricular heterotopia	0.1%	2
Polycythemia vera	0.1%	2
Post Polio syndrome	0.1%	2
Pseudotumor cerebri	0.1%	2
Reducing body myopathy	0.1%	2
Relapsing polychondritis	0.1%	2
Rubinstein-Taybi syndrome	0.1%	2
Sickle beta thalassemia	0.1%	2
Soft tissue sarcoma	0.1%	2
Spinal muscular atrophy 1	0.1%	2
Spinal muscular atrophy with respiratory distress 1	0.1%	2
Spinocerebellar ataxia	0.1%	2
Urea cycle disorders	0.1%	2
Waldenstrom macroglobulinemia	0.1%	2
WHIM syndrome	0.1%	2
Wilson disease	0.1%	2
22q11.2 duplication syndrome	0.1%	1
22q13.3 deletion syndrome	0.1%	1
2q23.1 microdeletion syndrome	0.1%	1
48,XXYY syndrome	0.1%	1
5q14.3 microdeletion syndrome	0.1%	1
6-pyruvoyl-tetrahydropterin synthase deficiency	0.1%	1
Aarskog syndrome	0.1%	1
Acquired Von Willebrand syndrome	0.1%	1
Acute intermittent porphyria	0.1%	1

	All Rare Caregivers (n=1,406)	
	Percentage	Number
Acute necrotizing encephalopathy	0.1%	1
Addison's disease	0.1%	1
ADNP syndrome	0.1%	1
Aganglionosis, total intestinal	0.1%	1
Alexander disease	0.1%	1
ALK+ histiocytosis	0.1%	1
Alpha-thalassemia	0.1%	1
Alport syndrome	0.1%	1
Alström syndrome	0.1%	1
Alzheimer's disease without neurofibrillary tangles	0.1%	1
Amyotrophic lateral sclerosis	0.1%	1
Anaplastic astrocytoma	0.1%	1
Angelman syndrome	0.1%	1
Aortic coarctation	0.1%	1
Arginase deficiency	0.1%	1
Arterial calcification of infancy	0.1%	1
Aspartylglycosaminuria	0.1%	1
Autoimmune hepatitis	0.1%	1
Autoimmune lymphoproliferative syndrome	0.1%	1
Autosomal dominant intermediate Charcot-Marie-Tooth	0.1%	1
Autosomal dominant leukodystrophy with autonomic	0.1%	1
Autosomal recessive polycystic kidney disease	0.1%	1
Axenfeld-Rieger syndrome	0.1%	1
Barth syndrome	0.1%	1
Becker muscular dystrophy	0.1%	1
Beta-Propeller Protein-Associated Neurodegeneration	0.1%	1
Bilateral perisylvian polymicrogyria	0.1%	1
Blepharophimosis intellectual disability syndromes	0.1%	1
Blepharophimosis, ptosis, and epicanthus inversus	0.1%	1
Borjeson-Forssman-Lehmann syndrome	0.1%	1
Budd-Chiari syndrome	0.1%	1
Cabezas syndrome	0.1%	1
CASK-Related Disorders	0.1%	1
Caudal regression syndrome	0.1%	1
Celiac artery compression syndrome	0.1%	1
Cerebral palsy athetoid	0.1%	1
Cerebro-facio-articular syndrome	0.1%	1
Children's interstitial lung disease	0.1%	1
Chordoma	0.1%	1

	All Rare Caregivers (n=1,406)	
	Percentage	Number
Chromosome 11q deletion	0.1%	1
Chromosome 12p deletion	0.1%	1
Chromosome 12q duplication	0.1%	1
Chromosome 15q duplication	0.1%	1
Chromosome 19q duplication	0.1%	1
Chromosome 1p36 deletion syndrome	0.1%	1
Chromosome 21q deletion	0.1%	1
Chromosome 2q deletion	0.1%	1
Chromosome 3p deletion	0.1%	1
Chromosome 8p duplication	0.1%	1
Chromosome 8q deletion	0.1%	1
Chromosome 8q24.3 deletion syndrome	0.1%	1
Chronic graft versus host disease	0.1%	1
Chronic granulomatous disease	0.1%	1
Chronic recurrent multifocal osteomyelitis	0.1%	1
Congenital contractural arachnodactyly	0.1%	1
Congenital fiber type disproportion	0.1%	1
Congenital human immunodeficiency virus	0.1%	1
Congenital sucrase-isomaltase deficiency	0.1%	1
Cowden syndrome	0.1%	1
Cri du chat syndrome	0.1%	1
Cryopyrin-associated periodic syndrome	0.1%	1
Cushing's syndrome	0.1%	1
Cyclic vomiting syndrome	0.1%	1
Cystic adenomatoid malformation of lung	0.1%	1
Cystinuria	0.1%	1
Diabetic mastopathy	0.1%	1
Diffuse cutaneous systemic sclerosis	0.1%	1
Dilated cardiomyopathy	0.1%	1
Disorders of Intracellular Cobalamin Metabolism	0.1%	1
Dopa-responsive dystonia	0.1%	1
Duane syndrome type 3	0.1%	1
Dysautonomia like disorder	0.1%	1
Dysfibrinogenemia	0.1%	1
Early infantile epileptic encephalopathy 25	0.1%	1
Early-onset generalized dystonia	0.1%	1
Eosinophilic cryptitis	0.1%	1
Erdheim-Chester disease	0.1%	1
Esophageal varices	0.1%	1

	All Rare Caregivers (n=1,406)	
	Percentage	Number
Familial adenomatous polyposis	0.1%	1
Familial Mediterranean fever	0.1%	1
Familial neurocardiogenic syncope	0.1%	1
Familial partial lipodystrophy	0.1%	1
Familial partial lipodystrophy associated with PPARG	0.1%	1
Familial primary hypomagnesemia	0.1%	1
Feingold syndrome	0.1%	1
Fibro-adipose vascular anomaly	0.1%	1
Fibrodysplasia ossificans progressiva	0.1%	1
Fragile X syndrome	0.1%	1
Freeman Sheldon syndrome	0.1%	1
Gaucher disease type 3	0.1%	1
Genetic lipodystrophy	0.1%	1
Giant platelet syndrome	0.1%	1
Glaucoma 3 primary infantile B	0.1%	1
Glioblastoma	0.1%	1
Glucose transporter type 1 deficiency syndrome	0.1%	1
Goldenhar disease	0.1%	1
Gorham's disease	0.1%	1
Granulomatosis with polyangiitis	0.1%	1
Growth hormone deficiency	0.1%	1
Hemolytic uremic syndrome	0.1%	1
Henoch-Schonlein purpura	0.1%	1
Hereditary sensory and autonomic neuropathy type 2	0.1%	1
Heterotaxy	0.1%	1
Holoprosencephaly	0.1%	1
Holzgreve syndrome	0.1%	1
Homocystinuria due to MTHFR deficiency	0.1%	1
Hydranencephaly	0.1%	1
Hydrocephalus	0.1%	1
Hyper-IgD syndrome	0.1%	1
Hyperinsulinism, diffuse	0.1%	1
I cell disease	0.1%	1
Ichthyosis vulgaris	0.1%	1
Inclusion body myopathy 2	0.1%	1
Inclusion body myositis	0.1%	1
Incontinentia pigmenti	0.1%	1
Infantile-onset ascending hereditary spastic paralysis	0.1%	1
Intellectual disability-severe speech delay-mild	0.1%	1

	All Rare Caregivers (n=1,406)	
	Percentage	Number
Intestinal lymphangiectasia	0.1%	1
Intracranial arteriovenous malformation	0.1%	1
Intrahepatic cholangiocarcinoma	0.1%	1
Isolated growth hormone deficiency	0.1%	1
Jacobsen syndrome	0.1%	1
Juvenile dermatomyositis	0.1%	1
Kabuki syndrome	0.1%	1
KBG syndrome	0.1%	1
Kleefstra syndrome	0.1%	1
Koolen de Vries syndrome	0.1%	1
L-2-hydroxyglutaric aciduria	0.1%	1
Lambdoid synostosis	0.1%	1
Langerhans cell histiocytosis	0.1%	1
Leukoencephalopathy with vanishing white matter	0.1%	1
Leukoencephalopathy, cerebral calcifications, and cysts	0.1%	1
Linear scleroderma	0.1%	1
LIPE-related familial partial lipodystrophy	0.1%	1
Lung adenocarcinoma	0.1%	1
Lupus nephritis	0.1%	1
Lymphangiomatosis	0.1%	1
Macrocephaly mesodermal hamartoma spectrum	0.1%	1
Macrocephaly-capillary malformation	0.1%	1
Maple syrup urine disease	0.1%	1
Marden-Walker syndrome	0.1%	1
Marinesco-Sjogren syndrome	0.1%	1
Mast cell activation syndrome	0.1%	1
Methylmalonic acidemia	0.1%	1
Methylmalonic aciduria, cblA type	0.1%	1
Microcephaly, holoprosencephaly, and intrauterine growth	0.1%	1
Microphthalmia microtia fetal akinesia	0.1%	1
Miller-Dieker syndrome	0.1%	1
Mosaic trisomy 22	0.1%	1
Mowat-Wilson syndrome	0.1%	1
Mucopolysaccharidosis type IIIB	0.1%	1
Mucopolysaccharidosis type IVA	0.1%	1
Myopathy congenital	0.1%	1
Nail-patella syndrome	0.1%	1
Neonatal adrenoleukodystrophy	0.1%	1
Nephrogenic diabetes insipidus	0.1%	1

	All Rare Caregivers (n=1,406)	
	Percentage	Number
Nephropathic cystinosis	0.1%	1
Netherton syndrome	0.1%	1
Neuroblastoma	0.1%	1
Neurofibroma	0.1%	1
Neurofibromatosis	0.1%	1
Neurofibromatosis type 1	0.1%	1
Oculodentodigital dysplasia	0.1%	1
Omenn syndrome	0.1%	1
Osteogenesis imperfecta type VI	0.1%	1
Osteogenesis imperfecta type VIII	0.1%	1
Pallister-Killian mosaic syndrome	0.1%	1
Paraneoplastic cerebellar degeneration	0.1%	1
Paroxysmal nocturnal hemoglobinuria	0.1%	1
PCDH19-related female-limited epilepsy	0.1%	1
Pelizaeus-Merzbacher disease	0.1%	1
Peripheral T-cell lymphoma	0.1%	1
Pheochromocytoma	0.1%	1
Pigmented villonodular synovitis	0.1%	1
Pilocytic astrocytoma	0.1%	1
Pitt-Hopkins syndrome	0.1%	1
Polyglucosan body disease, adult	0.1%	1
Primary hyperoxaluria type 1	0.1%	1
Progressive pseudorheumatoid dysplasia	0.1%	1
Progressive supranuclear palsy	0.1%	1
Pseudomyxoma peritonei	0.1%	1
Pseudo-Von Willebrand disease	0.1%	1
PURA syndrome	0.1%	1
Pyruvate carboxylase deficiency	0.1%	1
Rasmussen encephalitis	0.1%	1
Renal cell carcinoma	0.1%	1
Retinopathy, arteriosclerotic	0.1%	1
Rheumatic Fever	0.1%	1
Rhizomelic chondrodysplasia punctata type 1	0.1%	1
Rickets	0.1%	1
Ring chromosome 15	0.1%	1
Russell-Silver syndrome	0.1%	1
Schinzel Giedion syndrome	0.1%	1
Schizencephaly	0.1%	1
Severe congenital neutropenia autosomal dominant	0.1%	1

	All Rare Caregivers (n=1,406)	
	Percentage	Number
Sheehan syndrome	**0.1%**	1
Sickle cell disease associated with an other hemoglobin	**0.1%**	1
Snyder-Robinson syndrome	**0.1%**	1
Spinocerebellar ataxia 17	**0.1%**	1
Spinocerebellar ataxia 2	**0.1%**	1
Spinocerebellar ataxia 20	**0.1%**	1
Spinocerebellar ataxia 5	**0.1%**	1
Spondylothoracic dysostosis	**0.1%**	1
Stargardt disease	**0.1%**	1
Subcortical band heterotopia	**0.1%**	1
Succinic semialdehyde dehydrogenase deficiency	**0.1%**	1
Systemic scleroderma	**0.1%**	1
T cell immunodeficiency primary	**0.1%**	1
Tetrasomy X	**0.1%**	1
Timothy syndrome	**0.1%**	1
Trichorhinophalangeal syndrome type 2	**0.1%**	1
Trichothiodystrophy	**0.1%**	1
Tumefactive multiple sclerosis	**0.1%**	1
Turcot syndrome	**0.1%**	1
Turner syndrome	**0.1%**	1
Von Hippel-Lindau disease	**0.1%**	1
Wolf-Hirschhorn syndrome	**0.1%**	1
X-linked adrenoleukodystrophy	**0.1%**	1
X-linked severe combined immunodeficiency	**0.1%**	1
Other – not listed on GARD list	**6.5%**	92
Unknown or Undiagnosed	**2.3%**	33