List of the 30 most frequent Confirmed Rare Disease Diagnoses

ORPHA code	Disease name	Percent of Total Frequency	Preferential Parent
636	Neurofibromatosis type 1	6,9	Rare developmental defect during embryogenesis
145	Hereditary breast and ovarian cancer syndrome	4,6	Rare neoplastic disease
285	Ehlers-Danlos syndrome, hypermobility type	4,2	Rare systemic or rheumatologic disease
870	Down syndrome	4,0	Rare developmental defect during embryogenesis
399	Huntington disease	2,0	Rare neurologic disease
648	Noonan syndrome	1,8	Rare developmental defect during embryogenesis
758	Pseudoxanthoma elasticum	1,5	Rare systemic or rheumatologic disease
567	22q11.2 deletion syndrome	1,5	Rare developmental defect during embryogenesis
144	Lynch syndrome	1,2	Rare neoplastic disease
558	Marfan syndrome	1,2	Rare systemic or rheumatologic disease
98053	Rare genetic disease	1,2	Group of disorders
908	Fragile X syndrome	1,2	Rare developmental defect during embryogenesis
730	Autosomal dominant polycystic kidney disease	0,9	Rare renal disease
774	Hereditary hemorrhagic telangiectasia	0,9	Rare developmental defect during embryogenesis
68335	Chromosomal anomaly	0,8	Group of disorders
63	Alport syndrome	0,7	Rare renal disease
778	Rett syndrome	0,7	Rare neurologic disease
1727	22q11.2 microduplication syndrome	0,7	Rare developmental defect during embryogenesis
227535	Hereditary breast cancer	0,7	Rare neoplastic disease
138	CHARGE syndrome	0,6	Rare developmental defect during embryogenesis
321	Multiple osteochondromas	0,6	Rare bone disease
324	Fabry disease	0,6	Rare inborn errors of metabolism
610	Bethlem myopathy	0,6	Rare neurologic disease
739	Prader-Willi syndrome	0,6	Rare developmental defect during embryogenesis
821	Sotos syndrome	0,6	Rare developmental defect during embryogenesis
1906	Fetal valproate syndrome	0,6	Rare developmental defect during embryogenesis
2322	Kabuki syndrome	0,6	Rare developmental defect during embryogenesis
101081	Charcot-Marie-Tooth disease type 1A	0,6	Rare neurologic disease
101685	Rare non-syndromic intellectual disability	0,6	Rare neurologic disease
250989	1q21.1 microdeletion syndrome	0,6	Rare developmental defect during embryogenesis