

Table 1: Characteristics of the individuals with rare and unknown disease of the 522 RUDs reference sites involved in this study between 2019 and 2020

	All		Children		Adults	
	2019	2020	2019	2020	2019	2020
Patients (n)	145056	132282	58087	56124	85076	73839
Sex Ratio (M/F)	0.98	0.99	1.17	1.17	0.86	0.87
Number of visits (n)	300019	263380	122155	116347	175295	143565
Age (years)	25 [10, 55]	22 [9, 52]	8 [4, 13]	8 [3, 12]	50 [32, 66]	49 [32, 65]
Number of new confirmed diagnosis (n)	16799	8949	8306	5113	8494	3837
Levels of Diagnostics (n)						
Confirmed	223012	188304	82748	75562	139513	111578
Ongoing confirmation	34104	36468	16294	19398	16897	15696
Probable	20065	19910	9110	10000	10699	9624
Unknown	19830	15623	12438	9821	6755	5177
Missing	3008	3075	1565	1566	1431	1490

Table 2: Wilcoxon Tests to compare 2019 and 2020 activities in the BNDMR cohort

Test 2019 vs 2020	N 2019	N 2020	2019 Median [IQR]	2020 Median [IQR]	p-value
A. All activities					
Full cohort	300019	263380	223 [51, 647]	183.5 [55, 597.5]	0.36
Adult Patients	175431	143654	101 [22, 383.5]	87 [18, 298.5]	0.14
Child Patients	122019	116258	65 [13, 305]	54 [13, 275]	0.68
Lockdown Periods	85995	33987	64 [15, 189.25]	23.5 [8, 82.75]	6e-15
Not Lockdown Periods	214024	229393	157 [35.25, 464.5]	156.5 [47.25, 527]	0.28
Winter	65041	68875	45.5 [10, 138.75]	47 [12.25, 154.5]	0.14
First Lockdown	46019	25444	33 [8, 105.25]	19 [6, 59]	8.8e-06
Deconfinement	49399	44192	37.5 [9, 112.75]	34 [10, 119]	0.76
Summer break	36973	32930	28 [7, 91]	24 [7, 80.5]	0.39
Fall	53170	47733	39.5 [10, 123]	38 [11, 116]	0.94
Second Lockdown	39942	35663	31 [8, 93]	33 [9, 95.75]	0.76
Christmas break	9475	8543	9 [3, 24]	9 [3, 27]	0.5
B. All face-to-face activities					
Full cohort	292634	235534	217.5 [51, 638.75]	170 [50, 545.25]	0.088
Adult Patients	172237	127842	100.5 [22, 384.5]	72 [16, 284.5]	0.027
Child Patients	118015	104618	64 [13, 304]	54 [12.75, 259]	0.47
Lockdown Periods	84106	21960	64 [15, 179.75]	16.5 [6, 51.5]	<2e-16
Not Lockdown Periods	208528	213574	155 [35.25, 459.75]	153.5 [45, 481]	0.51
Winter	63298	66963	45 [10, 138.75]	47 [12, 145.75]	0.15
First Lockdown	44854	13917	33 [8, 103.25]	10 [4, 31.25]	<2e-16
Deconfinement	48146	37257	37.5 [9, 109.25]	30 [8.5, 93.5]	0.096
Summer break	35964	31093	28 [7, 89]	24 [7, 75.25]	0.3
Fall	51906	45225	39.5 [10, 123]	37 [10.25, 110]	0.79
Second Lockdown	39222	33036	30 [8, 90.5]	30 [8.75, 89]	0.79
Christmas break	9244	8043	9 [3, 24]	9 [3, 25]	0.79
C. Telehealth activities					
Full cohort	7385	27846	2 [1, 14]	19 [4, 72.25]	5e-14
Adult Patients	3194	15812	2 [1, 11.25]	12 [3, 43]	3e-09
Child Patients	4004	11640	2 [1, 14.25]	9 [2, 44.5]	2.7e-06
Lockdown Periods	1889	12027	2 [1, 13]	13 [3, 39]	5.1e-07
Not Lockdown Periods	5496	15819	2 [1, 14.25]	11 [3, 44.5]	8.6e-09
Winter	1743	1912	3 [1, 16]	2 [1, 7]	0.57
First Lockdown	1165	11527	3 [1, 12]	13 [3, 38.75]	2.1e-06
Deconfinement	1253	6935	2 [1, 16]	7 [3, 26]	0.0023
Summer break	1009	1837	3.5 [1, 25]	3 [2, 10]	0.53
Fall	1264	2508	3 [1, 28.5]	4 [2, 11]	0.83
Second Lockdown	720	2627	2 [1, 14.5]	4 [2, 14]	0.027
Christmas break	231	500	1 [1, 6]	2 [1, 5]	0.64

Table 3: Wilcoxon Tests to compare 2019 and 2020 activities by context in the BNDMR cohort

Test 2019 vs 2020	N 2019	N 2020	2019 Median [IQR]	2020 Median [IQR]	p-value
Consultation	194328	154348	143 [32, 417.25]	115 [28.5, 319]	0.066
Day Hospital	48217	38520	23 [5, 90.75]	17.5 [4, 71.25]	0.14
Conventional Hospitalization	25447	19498	16 [4, 65]	14 [4, 47]	0.19
Pluridisciplinary consultations	24642	23168	16 [3, 74]	14.5 [3, 68.25]	0.76
Telehealth activities	7385	27846	2 [1, 14]	19 [4, 72.25]	5e-14

Table 4: Numbers of activities by specialty network in France

Network name	Description	2019	2020
AnDDI	Developmental abnormalities, malformations and intellectual disability	31575	30971
BRAIN TEAM	Central nervous system diseases	6976	6695
Cardiogen	Hereditary cardiac diseases	8745	8468
DefiScience	Rare diseases affecting Cognitive development and causing intellectual disability	14712	15089
FAIIR	Autoimmune and autoinflammatory diseases	20819	20638
FAVA-Multi	Marfan Syndrome	4944	4527
FILFOIE	Rare liver diseaes	9399	9387
FILNEMUS	Neuromuscular diseases	38920	30683
FILSLAN	Amyotrophic lateral sclerosis	9650	8684
FIMARAD	Skin diseases	19642	15721
FIMATHO	Rare thoracic diseases	3902	3111
FIREENDO	Endocrinology	24085	16245
G2M	Metabolic diseases	14874	13738
MARIH	Immuno-hemateologic diseases	10767	8389
MCGRE	Rare red blood cells diseases	6035	6948
MHEMO	Constitutional haemorrhagic diseases	4880	5787
MUCO	Cystic fibrosis	578	569
NEUROSPHINX	Rare pelvic and spinal cord malformations	1647	560
ORKID	Kidney Diseases	32530	28624
OSCAR	Bones, Calcium and cartilage diseases	6461	6049
RESPIFIL	Respiratory diseases	2377	2161
SENSGENE	Rare sensory diseases	9319	7564
TETE COU	Head, Teeth and Neck diseases	17182	12772