

**Supplementary Table 2.** Total volume of activities prompted by ongoing care, CMA and WGS

Specialist	Ongoing Care (n=101)		CMA-/WGS- (n=71)		CMA-/WGS+ (n=22)	
Allergy & Immunology	5	1.2%	0	0.0%	1	3.8%
Cardiology	24	5.6%	0	0.0%	4	15.4%
Dentistry	7	1.6%	0	0.0%	0	0.0%
Dermatology	6	1.4%	0	0.0%	0	0.0%
Developmental Assessment	9	2.1%	0	0.0%	1	3.8%
Endocrinology	17	4.0%	0	0.0%	0	0.0%
ENT	21	4.9%	0	0.0%	0	0.0%
Gastroenterology	16	3.7%	0	0.0%	4	15.4%
Genetics	89	20.8%	0	0.0%	2	7.7%
Gynecology	2	0.5%	0	0.0%	0	0.0%
Hematology	9	2.1%	0	0.0%	0	0.0%
Metabolics	4	0.9%	0	0.0%	0	0.0%
MFM/Perinatology/Neonatology	7	1.6%	0	0.0%	0	0.0%
Nephrology	9	2.1%	0	0.0%	0	0.0%
Neurology	44	10.3%	0	0.0%	2	7.7%
Neurosurgery	13	3.0%	0	0.0%	1	3.8%
Oncology	2	0.5%	0	0.0%	1	3.8%
Ophthalmology/Optomety	39	9.1%	0	0.0%	5	19.2%
Orthopedics	23	5.4%	0	0.0%	1	3.8%
Pharmacology	0	0.0%	0	0.0%	0	0.0%
Plastic/General Surgery	20	4.7%	0	0.0%	0	0.0%
Psychiatry	5	1.2%	0	0.0%	0	0.0%
Pulmonology	6	1.4%	0	0.0%	1	3.8%
Rheumatology	5	1.2%	0	0.0%	0	0.0%
Urology	9	2.1%	0	0.0%	0	0.0%
Complex Care	8	1.9%	0	0.0%	0	0.0%
Palliative Care	1	0.2%	0	0.0%	0	0.0%
Sleep Clinic	8	1.9%	0	0.0%	3	11.5%
Respiratory Medicine	12	2.8%	0	0.0%	0	0.0%
Adolescent Medicine	2	0.5%	0	0.0%	0	0.0%
Anaesthesia	2	0.5%	0	0.0%	0	0.0%
Other	3	0.7%	0	0.0%	0	0.0%
Sub-total	427		0		26	
<b>Allied health</b>						
Audiology	21	16.2%	0	0.0%	1	8.3%
Dietetics/Nutrition	17	13.1%	0	0.0%	2	16.7%
Occupational Therapy	20	15.4%	0	0.0%	3	25.0%
Physiotherapy	20	15.4%	0	0.0%	3	25.0%
Speech/Language	8	6.2%	0	0.0%	2	16.7%
Genetic Counselling	38	29.2%	0	0.0%	0	0.0%
Psychology	4	3.1%	0	0.0%	0	0.0%
Support Group	1	0.8%	0	0.0%	1	8.3%
Other	1	0.8%	0	0.0%	0	0.0%
Sub-total	130		0	0.0%	12	100.0%
<b>Laboratory</b>						
Genetic	24	88.9%	6	100%	0	0%

Biochemical	2	7.4%	0	0%	0	0%
Other	1	3.7%	0	0%	2	100%
Sub-total	27		6		2	
<b>Imaging</b>						
Skeletal Survey	4	2%	0	0.0%	0	0.0%
Bone Age	3	1%	0	0.0%	0	0.0%
MRS/4 or MRI	38	18%	0	0.0%	2	33.3%
Ultrasound	64	31%	0	0.0%	2	33.3%
CT	5	2%	0	0.0%	0	0.0%
Xray	50	24%	0	0.0%	0	0.0%
Echocardiogram	27	13%	1	100.0%	1	16.7%
Ophthalmology imaging	2	1%	0	0.0%	1	16.7%
Other	14	7%	0	0.0%	0	0.0%
Sub-total	207		1		6	