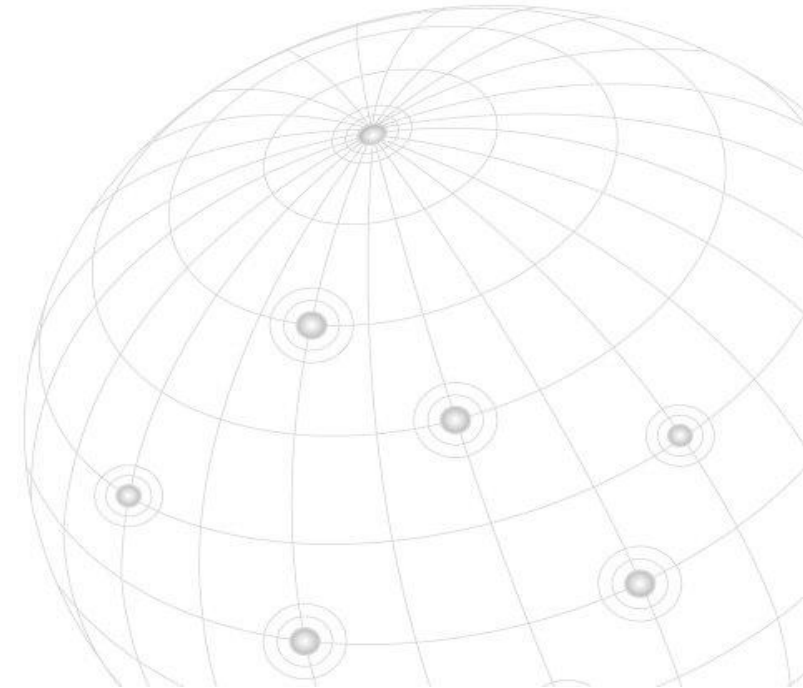




National Services Division
Germline Cancer Testing All Labs
22 October 2021



Germline Cancer Testing 2019-2020

- The following analysis is based on the data provided by the Labs for 2019-2020,
- The data was provided using a standard template format request,
- The data provided was not readily available or straightforward for Labs to access and provide and includes different interpretations from each Lab,
- The data is presented with assumptions and qualifications included,
- The data includes a further interpretation to consolidate where possible and provide a consistent and reliable analysis as far as possible with the information provided.

Germline Testing 2019-2020 Glasgow Samples In / Method Applied / Reports Out

#	Lab	INHERITED CANCER	Samples In	NGS	Sanger	MLPA	Reports Out
1	Glasgow	Birt Hogg Dubé syndrome	11		11		12
2	Glasgow	Colorectal cancer	0				
3	Glasgow	Cowden Syndrome/PTEN	11	11			24
4	Glasgow	DICER1 Syndrome	3	3			3
5	Glasgow	Glomus Tumours	0				
6	Glasgow	Gorlin Syndrome (Basal Cell Nevus Syndrome)	2	2			1
7	Glasgow	Hereditary Breast / Ovarian /Colorectal cancer syndrome	67	67			67
8	Glasgow	Hereditary Breast / Ovarian cancer syndrome	95	95			87
9	Glasgow	Hereditary breast cancer syndrome*	413	413			417
10	Glasgow	Hereditary Ovarian cancer syndrome	120	120			137
11	Glasgow	Hereditary Pancreatic Cancer	18	18			16
12	Glasgow	Juvenile Polyposis	0				
13	Glasgow	Li Fraumeni syndrome	22	22			22
14	Glasgow	Lynch Syndrome (HNPCC)	0				
15	Glasgow	Malignant Melanoma**	46	46			45
16	Glasgow	Medullary Thyroid cancer	0				
17	Glasgow	Neuroendocrine tumours	33	33			
18	Glasgow	Neurofibromatosis, NF1, schwannoma	78	65			
19	Glasgow	Peutz Jegher syndrome	0				
20	Glasgow	Polyposis	0				
21	Glasgow	Renal cancer	0				
22	Glasgow	Rhabdoid Tumour	1	1			0
		Totals	920	896	11	0	831

- Glasgow Raw Data,
- **During the course of this year, analysis for hereditary cancer changed from NGS analysis of two genes (and other genes on request) to an NGS subpanel of 8-22 genes depending on the referral reason. Before the subpanels were developed all referrals were tested under the test TS_BC so can not easily be categorised without checking each one individually. They are all included in the Hereditary breast cancer syndrome category,*
- ***During the course of this year, analysis for malignant melanoma changed from Sanger analysis of two genes to an NSG subpanel of 5 genes.*
- Does not include,
 - Samples received and stored,
 - 294 predictive tests carried out using Sanger analysis.

Germline Testing 2019-2020 Dundee Samples In / Method Applied / Reports Out

Lab	INHERITED CANCER	Samples In	NGS	Sanger	MLPA	Reports Out
1 Dundee	Birt Hogg Dubé syndrome	32		32		26
2 Dundee	colorectal cancer					
3 Dundee	Cowden Syndrome/PTEN					
4 Dundee	DICER1 SYNDROME					
5 Dundee	Glomus tumours	5	5			5
6 Dundee	GORLIN SYNDROME (BASAL CELL NEVUS SYNDROME)					
7 Dundee	Hereditary Breast / Ovarian /Colorectal cancer syndrome					
8 Dundee	hereditary breast / ovarian cancer syndrome					
9 Dundee	hereditary breast cancer syndrome					
10 Dundee	hereditary ovarian cancer syndrome					
11 Dundee	hereditary PANCREATIC CANCER					
12 Dundee	juvenile polyposis					
13 Dundee	li fraumeni syndrome					
14 Dundee	LYNCH SYNDROME (HNPCC)					
15 Dundee	malignant melanoma					
16 Dundee	MEDULLARY THYROID CANCER	6	6			6
17 Dundee	Neuroendocrine tumours	33	33			27
18 Dundee	Neurofibromatosis, NF1, schwannoma	78	65			65
19 Dundee	peutz jegher syndrome					
20 Dundee	polyposis					
21 Dundee	renal cancer	31	31			24
22 Dundee	RHABDOID TUMOUR					
	Totals	185	140	32		153

- Dundee Raw Data,
 - *does not include testing or reports for predictive/pre-symptomatic testing.*

Germline Testing 2019-2020 Aberdeen Samples In / Method Applied / Reports Out

Lab	INHERITED CANCER	Samples In	NGS	Sanger	MLPA	Reports Out
1 Aberdeen	Birt Hogg Dubé syndrome					
2 Aberdeen	Colorectal cancer					
3 Aberdeen	Cowden Syndrome/PTEN					
4 Aberdeen	DICER1 Syndrome					
5 Aberdeen	Glomus Tumours					
6 Aberdeen	Gorlin Syndrome (Basal Cell Nevus Syndrome)					
7 Aberdeen	Hereditary Breast / Ovarian /Colorectal cancer syndrome	232	236	56	64	236
8 Aberdeen	Hereditary Breast / Ovarian cancer syndrome	370	520	98	110	520
9 Aberdeen	Hereditary breast cancer syndrome*	191	204	126	71	258
10 Aberdeen	Hereditary Ovarian cancer syndrome	294	0	279	15	294
11 Aberdeen	Hereditary Pancreatic Cancer					
12 Aberdeen	Juvenile Polyposis					
13 Aberdeen	Li Fraumeni syndrome	20	5	13	13	18
14 Aberdeen	Lynch Syndrome (HNPCC)					
15 Aberdeen	Malignant Melanoma**					
16 Aberdeen	Medullary Thyroid cancer					
17 Aberdeen	Neuroendocrine tumours					
18 Aberdeen	Neurofibromatosis, NF1, schwannoma					
19 Aberdeen	Peutz Jegher syndrome					
20 Aberdeen	Polyposis					
21 Aberdeen	Renal cancer					
22 Aberdeen	Rhabdoid Tumour					
	Totals	1,107	965	572	273	1,326

- Aberdeen Raw Data,
 - *NGS Panels 2019-2020 spanned the period when we transferred the BRCA1/2 service to the expanded panel resulting in a number of samples (previously received) with strong family histories that were "reactivated" and tested using the expanded panel.*

Germline Testing 2019-2020 Edinburgh Samples In / Method Applied / Reports Out

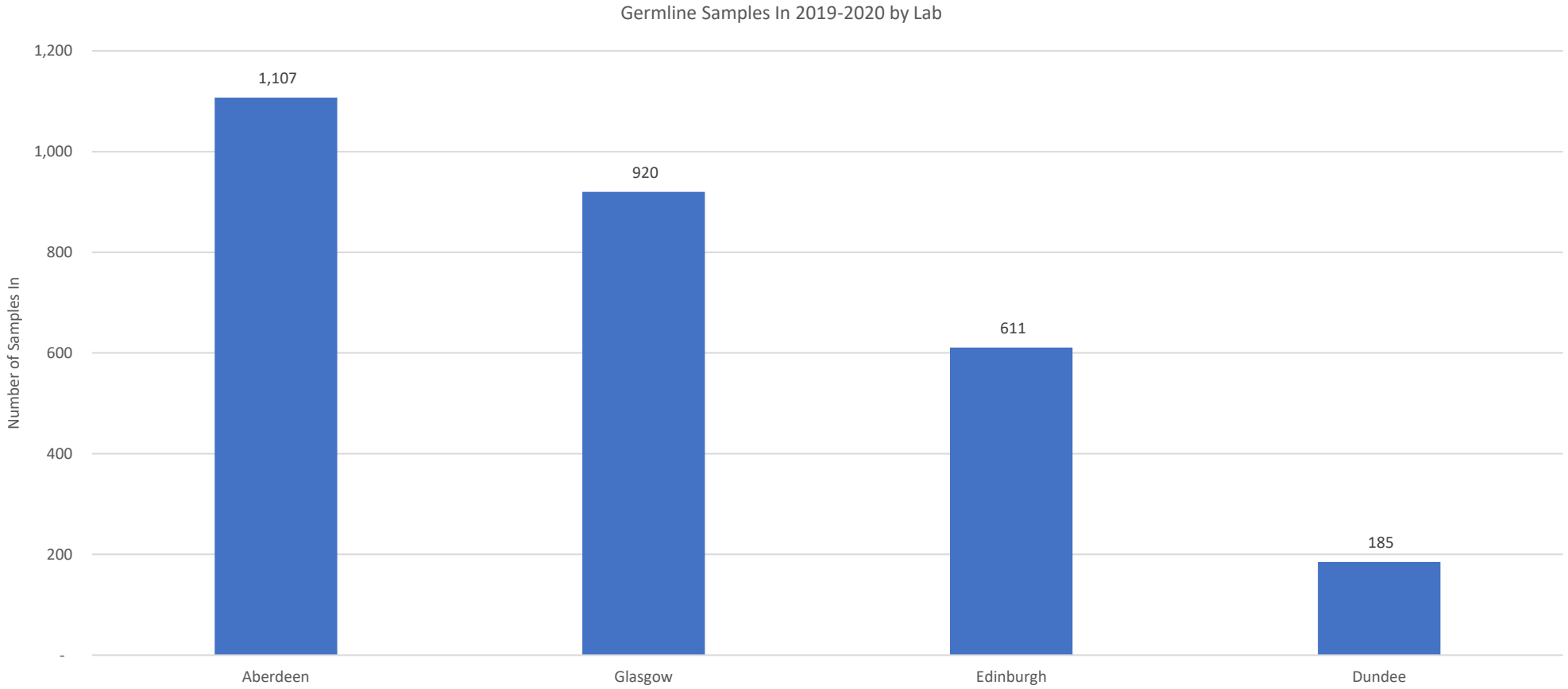
Lab	INHERITED CANCER	Samples In	NGS	Sanger	MILPA	Reports Out
1 Edinburgh	Birt Hogg Dubé syndrome					
2 Edinburgh	Colorectal cancer	136	136	136	136	136
3 Edinburgh	Cowden Syndrome/PTEN					
4 Edinburgh	DICER1 Syndrome	2	2	1		2
5 Edinburgh	Glomus Tumours	6		6		6
6 Edinburgh	Gorlin Syndrome (Basal Cell Nevus Syndrome)					
7 Edinburgh	Hereditary Breast / Ovarian /Colorectal cancer syndrome					
8 Edinburgh	Hereditary Breast / Ovarian cancer syndrome					
9 Edinburgh	Hereditary breast cancer syndrome*					
10 Edinburgh	Hereditary Ovarian cancer syndrome					
11 Edinburgh	Hereditary Pancreatic Cancer					
12 Edinburgh	Juvenile Polyposis	11	3	9	3	11
13 Edinburgh	Li Fraumeni syndrome					
14 Edinburgh	Lynch Syndrome (HNPCC)	361	184	324	221	361
15 Edinburgh	Malignant Melanoma**					
16 Edinburgh	Medullary Thyroid cancer					
17 Edinburgh	Neuroendocrine tumours					
18 Edinburgh	Neurofibromatosis, NF1, schwannoma					
19 Edinburgh	Peutz Jegher syndrome	6	6	1	6	6
20 Edinburgh	Polyposis	89	50	83	56	89
21 Edinburgh	Renal cancer					
22 Edinburgh	Rhabdoid Tumour					
	Totals	611	381	560	422	611

- Edinburgh Raw Data,
 - marked whether the sample has *received sanger, not the number of sanger.*

All Labs 2019-2020 Samples In

#	Sum of Samples In	Locations	Dundee	Edinburgh	Glasgow	Aberdeen	Totals
1	Birt Hogg Dubé syndrome	2	32		11		43
2	Colorectal cancer	1		136	-		136
3	Cowden Syndrome/PTEN	1			11		11
4	DICER1 Syndrome	2		2	3		5
5	Glomus Tumours	2	5	6	-		11
6	Gorlin Syndrome (Basal Cell Nevus Syndrome)	1			2		2
7	Hereditary Breast / Ovarian /Colorectal cancer syndrome	2			67	232	299
8	Hereditary Breast / Ovarian cancer syndrome	2			95	370	465
9	Hereditary breast cancer syndrome*	2			413	191	604
10	Hereditary Ovarian cancer syndrome	2			120	294	414
11	Hereditary Pancreatic Cancer	1			18		18
12	Juvenile Polyposis	1		11	-		11
13	Li Fraumeni syndrome	2			22	20	42
14	Lynch Syndrome (HNPCC)	1		361	-		361
15	Malignant Melanoma**	1			46		46
16	Medullary Thyroid cancer	1	6		-		6
17	Neuroendocrine tumours	2	33		33		66
18	Neurofibromatosis, NF1, schwannoma	2	78		78		156
19	Peutz Jegher syndrome	1		6	-		6
20	Polyposis	1		89	-		89
21	Renal cancer	1	31		-		31
22	Rhabdoid Tumour	1			1		1
	Totals		185	611	920	1,107	2,823

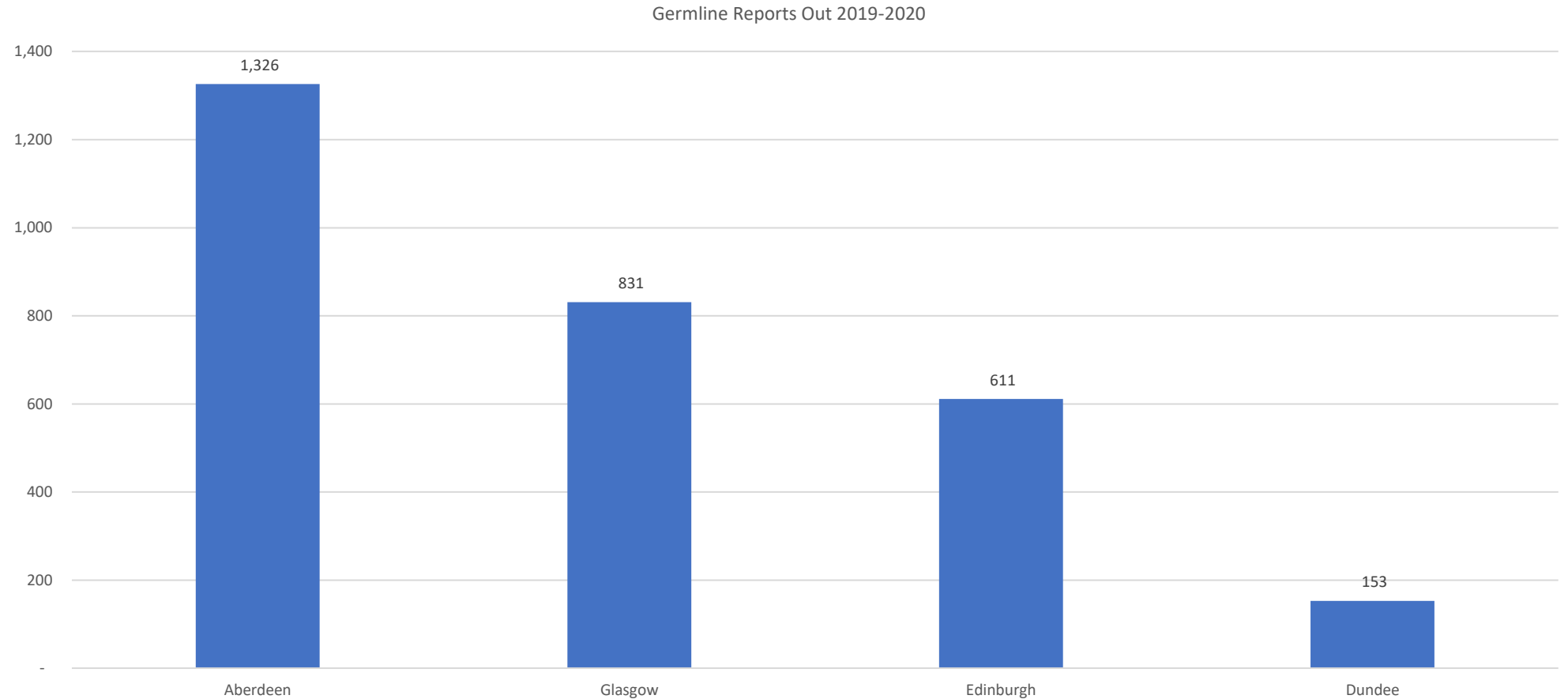
Germline 2019-2020 Samples In by Lab



All Labs 2019-2020 Reports Out

#	Sum of Reports Out	Locations	Dundee	Edinburgh	Glasgow	Aberdeen	Totals
1	Birt Hogg Dubé syndrome	2	26		12		38
2	Colorectal cancer	1		136			136
3	Cowden Syndrome/PTEN	1			24		24
4	DICER1 Syndrome	2		2	3		5
5	Glomus Tumours	2	5	6			11
6	Gorlin Syndrome (Basal Cell Nevus Syndrome)	1			1		1
7	Hereditary Breast / Ovarian /Colorectal cancer syndrome	2			67	236	303
8	Hereditary Breast / Ovarian cancer syndrome	2			87	520	607
9	Hereditary breast cancer syndrome*	2			417	258	675
10	Hereditary Ovarian cancer syndrome	2			137	294	431
11	Hereditary Pancreatic Cancer	1			16		16
12	Juvenile Polyposis	1		11			11
13	Li Fraumeni syndrome	2			22	18	40
14	Lynch Syndrome (HNPCC)	1		361			361
15	Malignant Melanoma**	1			45		45
16	Medullary Thyroid cancer	1	6				6
17	Neuroendocrine tumours	2	27				27
18	Neurofibromatosis, NF1, schwannoma	2	65				65
19	Peutz Jegher syndrome	1		6			6
20	Polyposis	1		89			89
21	Renal cancer	1	24				24
22	Rhabdoid Tumour	1			0		0
	Totals		153	611	831	1326	2921

All Labs 2019-2020 Germline Reports Out

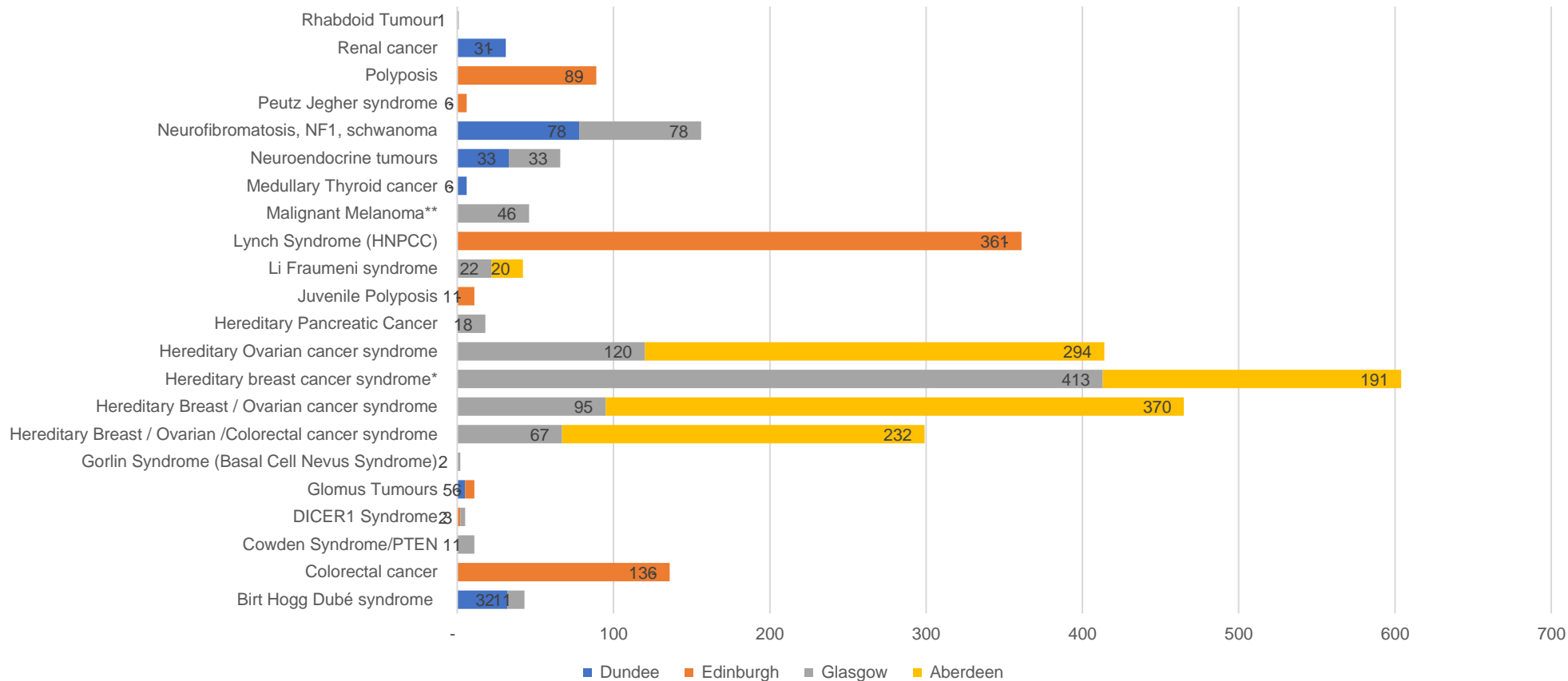


All Labs 2019-2020 Consolidated Samples / Method and Reports Out

INHERITED CANCER	Samples In	NGS	Sanger	MLPA	Reports Out
1Birt Hogg Dubé syndrome	43	-	-	43	38
2Colorectal cancer	136	136	136	136	136
3Cowden Syndrome/PTEN	11	11	-	-	24
4DICER1 Syndrome	5	5	1	-	5
5Glomus Tumours	11	5	6	-	11
6Gorlin Syndrome (Basal Cell Nevus Syndrome)	2	2	-	-	1
7Hereditary Breast / Ovarian /Colorectal cancer syndrome	299	303	56	64	303
8Hereditary Breast / Ovarian cancer syndrome	465	615	98	110	607
9Hereditary breast cancer syndrome*	604	617	126	71	675
10Hereditary Ovarian cancer syndrome	414	120	279	15	431
11Hereditary Pancreatic Cancer	18	18	-	-	16
12Juvenile Polyposis	11	3	9	3	11
13Li Fraumeni syndrome	42	27	13	13	40
14Lynch Syndrome (HNPCC)	361	184	324	221	361
15Malignant Melanoma**	46	46	-	-	45
16Medullary Thyroid cancer	6	6	-	-	6
17Neuroendocrine tumours	66	66	-	-	27
18Neurofibromatosis, NF1, schwannoma	156	130	-	-	65
19Peutz Jegher syndrome	6	6	1	6	6
20Polyposis	89	50	83	56	89
21Renal cancer	31	31	-	-	24
22Rhabdoid Tumour	1	1	-	-	-
Totals	2,823	2,382	1,175	695	2,921

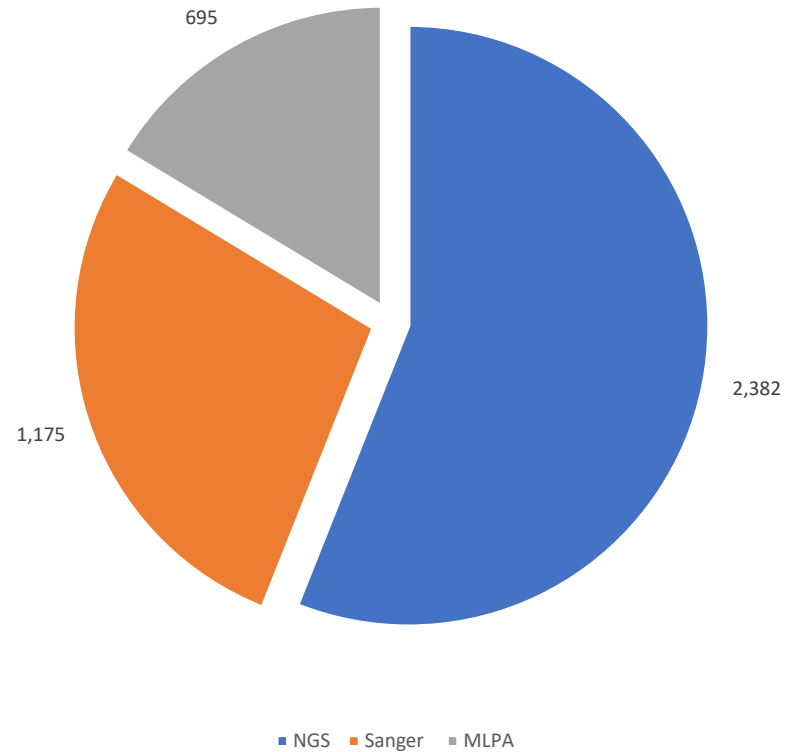
Germline Samples In by Lab by Disease Type

Germline Samples In by Lab 2019-2020



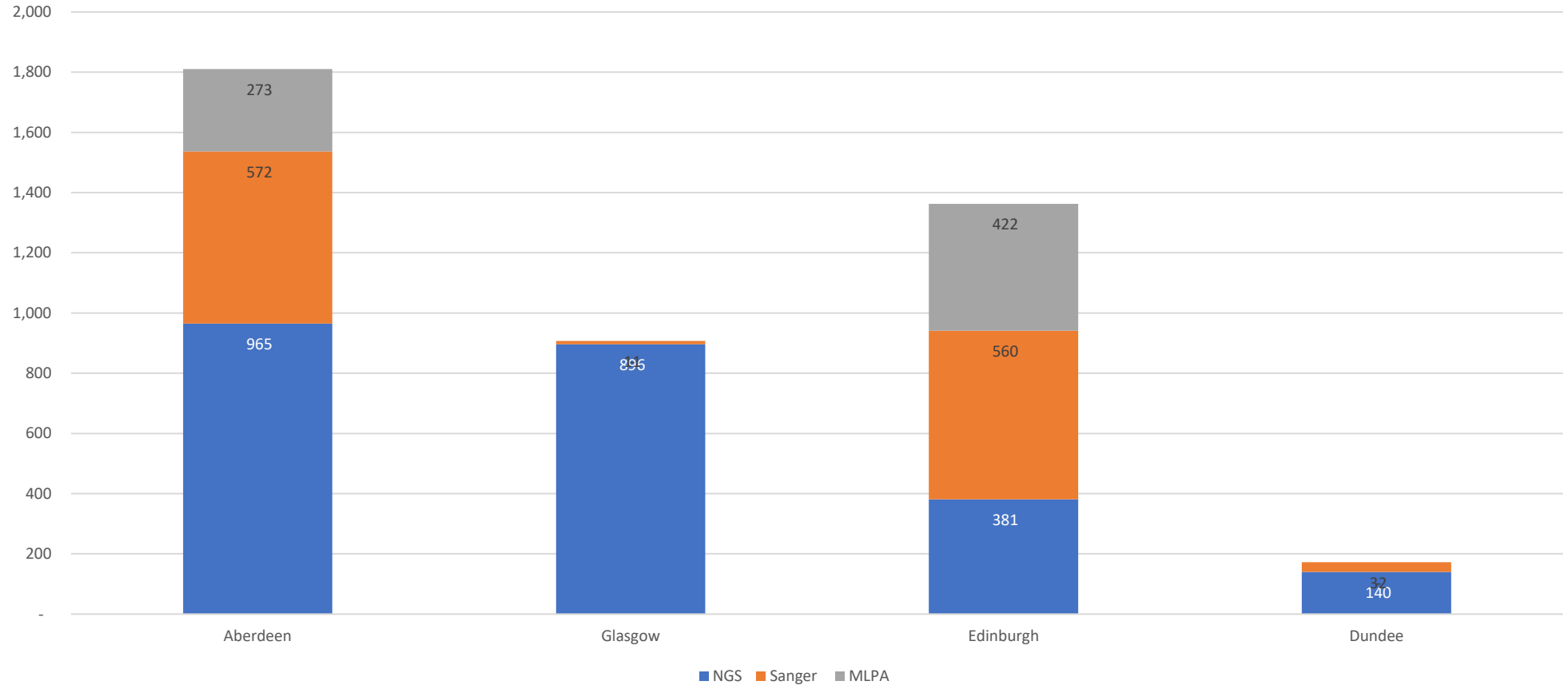
Germline 2019-2020 Testing Method is predominantly (85%) NGS

Germline Test Methods by Type 2019-2020 All Labs



Germline 2019-2020 Test Methods by Lab

Germline Test Methods 2019-2020



Summary

- This exercise for Germline testing highlight the same issue identified from the earlier Somatic analysis to identify workload and activity levels provided by the Consortia as the sum of the parts of each individual Lab,
- The number and types of test by test method – a fundamental issue in understanding existing and determining future capacity requirements – is not routinely collected or reported by the Labs,
- The number and types of test by Method is required to establish future investment requirements in people, equipment and technology, facilities and infrastructure to organise and manage and build on the existing capability to meet future increasing and different types of demand.