

# RESEARCH REPORT



## Genetic Testing Markets.

Global Market Analysis with Forecasts by Applications,  
Technologies, Product and User. With Executive and Consultant  
Guides.

2023 to 2027





Howe Sound Research is a market research and consulting company based in Vancouver, British Columbia, Canada. In our spare time we like to sail in a large body of water nearby called Howe Sound. We publish market research reports that address scientific industries with an emphasis on Biotechnology and Clinical Diagnostic markets. We consider ourselves experts in these areas.

We approach market research differently than other companies. At any one time we have a limited number of reports, and we update them frequently, sometimes several times a year. Our reports are prepared by people who understand the industry and have worked and studied in the area. This contrasts with the many research mills who produce canned reports on the Handbag market one day and the X-Ray market the next.

Our intellectual property is encryption protected. If you have issues with this or would like to upgrade to a corporate license that is not encrypted, please contact us at the email above.

Please feel free to contact us if you have any questions. We believe in customer service and are more than willing to provide after sales assistance and custom data.

All Rights Reserved. This document contains copyrighted intellectual property. No part of it may be circulated, copied, quoted, or otherwise reproduced without the approval of Howe Sound Research.

Please respect our intellectual property and do not distribute this document beyond the terms of your license.

## Table of Contents

1	Market Guides .....	24
1.1	Genetic Testing - Strategic Situation Analysis .....	25
1.2	Guide for Executives, Marketing, Sales and Business Development Staff .....	27
1.3	Guide for Management Consultants and Investment Advisors.....	29
2	Introduction and Market Definition .....	31
2.1	Genetic Testing Definition in This Report .....	32
2.2	The Genomics Revolution.....	33
2.3	Market Definition.....	35
2.3.1	Revenue Market Size.....	35
2.3.2	Newborn Screening .....	36
2.3.3	Non Invasise Pregnancy Testing.....	36
2.3.4	Predictive .....	36
2.3.5	Oncology .....	36
2.3.6	Direct to Consumer .....	36
2.3.7	Other Application.....	37
2.3.8	PCR .....	37
2.3.9	NGS.....	37
2.3.10	Cytogenetic.....	37
2.3.11	Other Technology .....	37
2.4	Methodology.....	38
2.4.1	Methodology.....	38

2.4.2	Sources .....	38
2.4.3	Authors .....	38
2.5	Perspective: Healthcare and the IVD Industry .....	40
2.5.1	Global Healthcare Spending .....	40
2.5.2	Spending on Diagnostics .....	41
2.5.3	Important Role of Insurance for Diagnostics .....	43
2.6	Chromosomes, Genes and Epigenetics .....	44
2.6.1	Chromosomes .....	44
2.6.2	Genes .....	46
2.6.3	Epigenetics .....	49
3	Market Overview.....	51
3.1	Players in a Dynamic Market.....	52
3.1.1	Academic Research Lab .....	53
3.1.2	Diagnostic Test Developer.....	53
3.1.3	Instrumentation Supplier .....	54
3.1.4	Chemical/Reagent Supplier.....	54
3.1.5	Pathology Supplier .....	54
3.1.6	Independent Clinical Laboratory .....	55
3.1.7	Public National/regional Laboratory .....	55
3.1.8	Hospital Laboratory .....	56
3.1.9	Physicians Office Lab (POLs) .....	56
3.1.10	Audit Body .....	57
3.1.11	Certification Body.....	57
3.2	Genetic Tests –Types, Examples and Discussion .....	59

3.2.1	Preimplantation Genetic Diagnosis- An Emerging Market .....	59
3.2.2	Prenatal Diagnosis – New Technologies Create Opportunity .....	60
3.2.3	Newborn Screening .....	60
3.2.4	Diagnostic Testing .....	61
3.2.5	Carrier Testing .....	61
3.2.6	Predictive and Presymptomatic Testing.....	62
3.2.7	Pharmacogenomics .....	62
3.2.8	Forensic Testing.....	63
3.2.9	Parental Testing .....	63
3.2.10	Ancestral Testing.....	63
3.3	Industry Structure.....	65
3.3.1	Hospital's Testing Share.....	65
3.3.2	Economies of Scale.....	65
3.3.3	Hospital vs. Central Lab .....	66
3.3.4	Physician Office Lab's.....	67
3.3.5	Physician's and POCT .....	67
4	Market Trends .....	68
4.1	Factors Driving Growth .....	69
4.1.1	Genetic Discoveries Creating New Diagnostic Markets.....	69
4.1.2	The Aging Effect .....	70
4.1.3	Pharmacogenomics Drives Further Growth.....	72
4.1.4	Oncology and Liquid Biopsy Enter New Era.....	73
4.1.5	Fertility Practice Growth drives market.....	73
4.1.6	Direct to Consumer begins to break out.....	73

4.2	Factors Limiting Growth .....	74
4.2.1	Increased Competition Lowers Price.....	74
4.2.2	Lower Costs.....	75
4.2.3	Testing usage analysis curtailing growth .....	75
4.2.4	Wellness has a downside .....	75
4.3	Instrumentation, Automation and Diagnostic Trends .....	77
4.3.1	Traditional Automation and Centralization.....	77
4.3.2	The New Automation, Decentralization and Point of Care.....	77
4.3.3	Instruments Key to Market Share.....	79
4.3.4	Bioinformatics Plays a Role .....	79
4.3.5	PCR Takes Command.....	80
4.3.6	Next Generation Sequencing Fuels a Revolution.....	81
4.3.7	NGS Impact on Pricing.....	82
4.3.8	Whole Genome Sequencing, A Brave New World .....	82
4.3.9	Companion Diagnostics Blurs Diagnosis and Treatment .....	83
4.3.10	Shifting Role of Diagnostics.....	84
5	Genetic Testing Recent Developments .....	86
5.1	Recent Developments.....	87
5.1.1	Importance of This Section .....	87
5.1.2	How to Use This Section.....	87
5.2	Genomics England, Aims to Sequence 100K Newborns .....	87
5.3	Juno Diagnostics NIPT With At-Home Sample Collection .....	90
5.4	Cardio Diagnostics Launches Genetic Cardio Risk Test.....	93
5.5	Universal Genetic Testing in Breast Cancer Further Supported.....	96

5.6	Aniling Gets CE-IVD Marks for Cancer Sequencing Tests .....	98
5.7	MyOme Lands Investment for WGS.....	99
5.8	Type 2 Diabetes GWAS Enables Risk Prediction .....	100
5.9	LetsGetChecked to Acquire Veritas Genetics.....	102
5.10	Guardant Health Plans New Comprehensive Assay.....	103
5.11	Tesis Labs Raises \$20M.....	105
5.12	Sema4: Sema4 Elements .....	106
5.13	Clarified Precision Medicine Acquires Interpares Biomedicine.....	106
5.14	Genetic Technologies to Acquire DTC Genetic Test Provider EasyDNA .....	107
5.15	Illumina, Belgium Genetic Centers Partner on WGS-Based Dx.....	108
5.16	NIH, Genomics England Launch Sequencing Studies Using Nanopore Technology	108
5.17	Progenity Revenues Fall on COVID-19 Effect.....	110
5.18	Natera Launches Tumor Genomic Profiling Assay .....	112
5.19	23andMe To Go Public .....	115
5.20	Guardant Health to Offer Tumor Tissue Sequencing.....	115
5.21	Ancestry Quits Health Offering.....	119
5.22	DTC Firm MyDNA Merges With Gene by Gene .....	124
5.23	Aetna Coverage for NIPT to Include All Pregnancies .....	124
5.24	Agios Pharmaceuticals, PerkinElmer Partner on Hereditary Anemia Genetic Testing 125	
5.25	Tumor, Germline Testing of Cancer Patients Can Give Discordant Results.....	126
6	Profiles of Key Companies.....	129
6.1	10x Genomics, Inc.....	130
6.2	23andME Inc.....	131



6.3	Abbott Laboratories.....	132
6.4	Adaptive Biotechnologies.....	134
6.5	Admera Health, LLC.....	136
6.6	Agilent/Dako .....	137
6.7	Akonni Biosystems .....	139
6.8	Amoy Diagnostics Co., Ltd.....	140
6.9	Ancestry.com LLC .....	141
6.10	Anchor Dx.....	143
6.11	Arc Bio .....	144
6.12	Arrayit Corporation .....	145
6.13	ARUP Laboratories.....	147
6.14	Aus Diagnostics .....	149
6.15	Baylor Miraca Genetics Laboratories .....	150
6.16	Beckman Coulter Diagnostics.....	151
6.17	Becton, Dickinson and Company .....	152
6.18	BGI Genomics Co. Ltd .....	154
6.19	Bioarray Genetics.....	156
6.20	Biocept, Inc.....	157
6.21	Biodesix Inc. ....	158
6.22	BioFluidica.....	159
6.23	BioGenex .....	160
6.24	Biolidics Ltd.....	162
6.25	bioMérieux Diagnostics.....	163
6.26	Bioneer Corporation .....	165

6.27	Bio-Rad Laboratories, Inc.....	167
6.28	Bio-Techne .....	169
6.29	C2i Genomics.....	171
6.30	Caris Molecular Diagnostics.....	172
6.31	CellMax Life .....	173
6.32	Centogene.....	174
6.33	Cepheid (Danaher) .....	176
6.34	Circulogene.....	178
6.35	Clinical Genomics.....	179
6.36	Color Genomics .....	180
6.37	Complete Genomics, Inc. – A BGI Company .....	181
6.38	Dante Labs.....	183
6.39	Datar Cancer Genetics Limited.....	185
6.40	Diasorin S.p.A.....	186
6.41	Epic Sciences.....	188
6.42	Epigenomics AG.....	189
6.43	Eurofins Scientific .....	190
6.44	Excellerate Bioscience.....	192
6.45	Fabric Genomics .....	193
6.46	Freenome.....	194
6.47	FUJIFILM Wako Diagnostics.....	195
6.48	Fujirebio.....	196
6.49	Fulgent Genetics .....	197
6.50	Gene by Gene, Ltd.....	198

6.51	Genedrive.....	200
6.52	GeneFirst Ltd.....	201
6.53	Genetron Holdings.....	202
6.54	Genomics England.....	203
6.55	Genomics Personalized Health (GPH).....	205
6.56	GenomOncology.....	206
6.57	Genzyme Corporation.....	207
6.58	Grifols.....	209
6.59	Guardant Health.....	210
6.60	Guardiome.....	211
6.61	HeiScreen.....	213
6.62	Helix.....	214
6.63	Helomics.....	216
6.64	Hologic.....	217
6.65	HTG Molecular Diagnostics.....	219
6.66	Human Longevity, Inc.....	220
6.67	iCellate.....	222
6.68	Illumina.....	223
6.69	Incell Dx.....	224
6.70	Inivata.....	225
6.71	Integrated Diagnostics.....	226
6.72	Invitae Corporation.....	227
6.73	Invivoscribe.....	228
6.74	Karius.....	229

6.75	Lunglife AI Inc .....	231
6.76	MacroGen.....	232
6.77	MDNA Life SCIENCES, Inc.....	233
6.78	MDx Health .....	234
6.79	Medgenome .....	235
6.80	Menarini Silicon Biosystems.....	236
6.81	Meridian Bioscience.....	237
6.82	Mesa Biotech (Thermo Fisher) .....	239
6.83	Mesa Laboratories, Inc. ....	240
6.84	miR Scientific.....	242
6.85	MNG Labs .....	243
6.86	NantHealth, Inc. ....	244
6.87	Natera .....	246
6.88	Nebula Genomics .....	247
6.89	NeoGenomics .....	248
6.90	New England Biolabs, Inc. ....	250
6.91	NGeneBio.....	251
6.92	Novogene Bioinformatics Technology Co., Ltd.....	253
6.93	Omega Bioservices .....	255
6.94	Oncocyte.....	257
6.95	OncoDNA .....	258
6.96	OpGen.....	259
6.97	ORIG3N, Inc. ....	260
6.98	Origene Technologies.....	262

6.99	Oxford Nanopore Technologies .....	263
6.100	Panagene.....	264
6.101	Perkin Elmer.....	265
6.102	Personal Genome Diagnostics.....	267
6.103	Personalis.....	268
6.104	Precipio .....	269
6.105	PrecisionMed .....	270
6.106	Promega .....	271
6.107	Protagen Diagnostics.....	273
6.108	Qiagen .....	274
6.109	QuantuMDx .....	276
6.110	Roche Molecular Diagnostics .....	277
6.111	Roswell Biotechnologies .....	279
6.112	Seegene.....	280
6.113	Sema4 Holdings.....	282
6.114	Sequencing.com.....	284
6.115	Siemens Healthineers .....	286
6.116	simfo GmbH .....	287
6.117	Singlera Genomics Inc.....	288
6.118	SkylineDx .....	289
6.119	Standard BioTools .....	290
6.120	Sure Genomics, Inc. ....	292
6.121	Sysmex.....	294
6.122	Sysmex Inostics.....	296

6.123	Tempus Labs, Inc. ....	297
6.124	Thermo Fisher Scientific Inc.....	298
6.125	Variantyx.....	301
6.126	Volition .....	302
6.127	Vyant Bio.....	303
7	Global Market Size .....	304
7.1	Global Market by Country .....	305
7.1.1	Table – Global Market by Country.....	305
7.1.2	Chart – Country Market Shares .....	306
7.2	Global Market by Application .....	307
7.2.1	Table – Global Market by Application .....	307
7.2.2	Chart – Application Share by Year .....	308
7.2.3	Chart – Application Segment Growth Rates .....	309
7.2.4	Chart – Application Segment Share Shifts.....	310
7.2.5	Chart – Application Segment Share Base Year .....	311
7.2.6	Chart – Application Segment Share Final Year .....	312
7.3	Global Market by Technology .....	313
7.3.1	Table – Global Market by Technology .....	313
7.3.2	Chart – Technology Share by Year .....	314
7.3.3	Chart – Technology Segment Growth Rates .....	315
7.3.4	Chart – Technology Segment Share Shifts.....	316
7.3.5	Chart – Technology Segment Share Base Year .....	317
7.3.6	Chart – Technology Segment Share Final Year .....	318
7.4	Global Market by Product.....	319

7.4.1	Table – Global Market by Product.....	319
7.4.2	Chart – Product Share by Year.....	320
7.4.3	Chart – Product Segment Growth Rates.....	321
7.4.4	Chart – Product Segment Share Shifts.....	322
7.4.5	Chart – Product Segment Share Base Year.....	323
7.4.6	Chart – Product Segment Share Final Year.....	324
7.5	Global Market by User.....	325
7.5.1	Table – Global Market by User.....	325
7.5.2	Chart – User Share by Year.....	326
7.5.3	Chart – User Segment Growth Rates.....	327
7.5.4	Chart – User Segment Share Shifts.....	328
7.5.5	Chart – User Segment Share Base Year.....	329
7.5.6	Chart – User Segment Share Final Year.....	330
8	Market Sizes by Application.....	331
8.1	Newborn Testing Market.....	332
8.1.1	Table Newborn – by Country.....	332
8.1.2	Chart – Newborn Growth.....	333
8.2	NIPT Market.....	334
8.2.1	Table NIPT – by Country.....	334
8.2.2	Chart – NIPT Growth.....	335
8.3	Predictive Testing Market.....	336
8.3.1	Table Predictive – by Country.....	336
8.3.2	Chart – Predictive Growth.....	337
8.4	Oncology Testing Market.....	338

8.4.1	Table Oncology – by Country .....	338
8.4.2	Chart – Oncology Growth .....	339
8.5	DTC Testing Market .....	340
8.5.1	Table DTC – by Country .....	340
8.5.2	Chart – DTC Growth .....	341
8.6	Other Testing Market.....	342
8.6.1	Table Other – by Country .....	342
8.6.2	Chart – Other Growth .....	343
9	Global Genetic Testing Market by Technology.....	344
9.1	PCR Testing Market .....	345
9.1.1	Table PCR – by Country .....	345
9.1.2	Chart – PCR Growth.....	346
9.2	NGS Market .....	347
9.2.1	Table NGS – by Country.....	347
9.2.2	Chart – NGS Growth.....	348
9.3	Cytogenetic Testing Market.....	349
9.3.1	Table Cytogenetic – by Country.....	349
9.3.2	Cytogenetic – Predictive Growth .....	350
9.4	Other Testing Market.....	351
9.4.1	Table Other – by Country .....	351
9.4.2	Chart – Other Growth .....	352
10	Global Genetic Testing Market by Product .....	353
10.1	Instruments Market .....	354
10.1.1	Table Instruments – by Country .....	354



10.1.2	Chart – Instrument Growth .....	355
10.2	Reagents and Kits Market.....	356
10.2.1	Table Reagents and Kits – by Country.....	356
10.2.2	Chart – Reagents and Kits Growth.....	357
10.3	Software Market .....	358
10.3.1	Table Software – by Country .....	358
10.3.2	Software Growth .....	359
10.4	Services Market .....	360
10.4.1	Table Services – by Country.....	360
10.4.2	Chart – Services Growth .....	361
11	Global Genetic Testing Market by User .....	362
11.1	Academic Market.....	363
11.1.1	Table Academic – by Country .....	363
11.1.2	Chart – Academic Growth.....	364
11.2	Clinical Market.....	365
11.2.1	Table Clinical – by Country.....	365
11.2.2	Chart – Clinical Growth.....	366
11.3	Pharmaceutical Market.....	367
11.3.1	Table Pharmaceutical – by Country .....	367
11.3.2	Pharmaceutical – Predictive Growth .....	368
11.4	Industrial and Other Market .....	369
11.4.1	Table Industrial and Other – by Country.....	369
11.4.2	Chart – Industrial and Other Growth .....	370
12	The Future of Genetic Testing .....	371

13 Appendices .....372

13.1 United States Medicare System: Laboratory Fees Schedule ..... 372

13.2 The Most Used IVD Assays..... 475

13.3 The Highest Grossing Assays ..... 480

## Table of Tables

Table 1 Most Common Traditional Genetic Disorders .....	33
Table 2 The Base Pairs.....	47
Table 3 Market Players by Type .....	53
Table 4 The Different Types of Genetic Tests .....	59
Table 5 Five Factors Driving Growth .....	69
Table 6 Four Factors Limiting Growth .....	74
Table 7 Seven Key Diagnostic Laboratory Technology Trends .....	77
Table 8 Global Market by Country.....	305
Table 9 Global Market by Application .....	307
Table 10 Global Market by Technology .....	313
Table 11 Global Market by Product.....	319
Table 12 Global Market by User .....	325
Table 13 Newborn Segment by Country .....	332
Table 14 NIPT Segment by Country .....	334
Table 15 Predictive Segment by Country .....	336
Table 16 Oncology Segment by Country.....	338
Table 17 DTC Segment by Country .....	340
Table 18 Other Segment by Country .....	342
Table 19 PCR Segment by Country .....	345
Table 20 NGS Segment by Country.....	347
Table 21 Cytogenetic Segment by Country .....	349

Table 22 Other Segment by Country .....	351
Table 23 Instruments Segment by Country .....	354
Table 24 Reagents and Kits Segment by Country .....	356
Table 25 Software Segment by Country .....	358
Table 26 Services Segment by Country .....	360
Table 27 Academic Segment by Country.....	363
Table 28 Clinical Segment by Country .....	365
Table 29 Pharmaceutical Segment by Country.....	367
Table 30 Industrial and Other Segment by Country .....	369
Table 31 Clinical Lab Fee Schedule .....	372
Table 32 The Most Common Assays.....	475
Table 33 Largest Revenue Assays.....	480

## Table of Figures

Figure 1 Global Health Care Spending.....	41
Figure 2 The Lab Test Pie .....	42
Figure 3 DNA Strands and Chromosomes .....	44
Figure 4 Karyogram of Human Chromosomes.....	45
Figure 5 Size of Various Genomes .....	48
Figure 6 The Road to Diagnostics .....	52
Figure 7 The Changing Age of the World Population.....	71
Figure 8 Health Care Consumption by Age .....	71
Figure 9 Cancer Incidence - Age at Diagnosis.....	72
Figure 10 Centralized vs. Decentralized Laboratory Service .....	78
Figure 11 A Highly Multiplexed Syndromic Testing Unit.....	78
Figure 12 The Real Cost to Sequence the Human Genome.....	81
Figure 13 The Codevelopment Process .....	83
Figure 14 Comparing MDx Diagnostic and Traditional Testing.....	85
Figure 15 Country Market Shares.....	306
Figure 16 Application Share by Year.....	308
Figure 17 Application Segment Growth Rates.....	309
Figure 18 Segment Share Shifts .....	310
Figure 19 Application Segment Share Base Year .....	311
Figure 20 Application Segment Share Final Year.....	312
Figure 21 Technology Share by Year.....	314

Figure 22 Technology Segment Growth Rates.....	315
Figure 23 Technology Segment Share Shifts .....	316
Figure 24 Technology Segment Share Base Year .....	317
Figure 25 Technology Segment Share Final Year.....	318
Figure 26 Product Share by Year .....	320
Figure 27 Product Segment Growth Rates .....	321
Figure 28 Product Segment Share Shifts.....	322
Figure 29 Product Segment Share Base Year .....	323
Figure 30 Product Segment Share Final Year .....	324
Figure 31 User Share by Year .....	326
Figure 32 User Segment Growth Rates.....	327
Figure 33 User Segment Share Shifts.....	328
Figure 34 User Segment Share Base Year .....	329
Figure 35 User Segment Share Final Year.....	330
Figure 36 Newborn vs. Total Market Growth .....	333
Figure 37 NIPT vs. Total Market Growth .....	335
Figure 38 Predictive vs. Total Market Growth .....	337
Figure 39 Oncology vs. Total Market Growth .....	339
Figure 40 DTC vs. Total Market Growth .....	341
Figure 41 Other vs. Total Market Growth .....	343
Figure 42 PCR vs. Total Market Growth .....	346
Figure 43 NGS vs. Total Market Growth.....	348
Figure 44 Cytogenetic vs. Total Market Growth.....	350
Figure 45 Other vs. Total Market Growth .....	352

Figure 46 Instrument vs. Total Market Growth..... 355

Figure 47 Reagents and Kits vs. Total Market Growth..... 357

Figure 48 Software vs. Total Market Growth ..... 359

Figure 49 Services vs. Total Market Growth ..... 361

Figure 50 Academic vs. Total Market Growth..... 364

Figure 51 Clinical vs. Total Market Growth..... 366

Figure 52 Pharmaceutical vs. Total Market Growth..... 368

Figure 53 Industrial and Other vs. Total Market Growth..... 370