A	genetic counseling
ACA. See Affordable Care Act	historical perspective, 83
AD. See Alzheimer's disease	overview, 73
ADA. See Americans with Disabilities Act	pretest counseling, 84-86
Adrenal carcinoma, genetic testing, 75	recent developments, 83-84
Affordable Care Act (ACA), 227	genetic testing
ALS. See Amyotrophic lateral sclerosis	biosample selection, 86
Alzheimer's disease (AD)	direct-to-consumer testing, 78
APOE testing, 49–50	germline testing, 87
direct-to-consumer testing, 49–50	guidelines, 76, 78–82
gene mutations, 48	indications, 74–76
genetic counseling, 48–49, 53	interpretation, 87–88
polygenic risk scores, 50	laboratory selection, 86
predictive testing, 48	polygenic risk scores, 87
Americans with Disabilities Act (ADA), 228	test selection, 86–87
Amyotrophic lateral sclerosis (ALS), genetic testing	pediatric considerations in testing and
and counseling, 51	counseling, 88–89
Angelman syndrome, 63	risk assessment tools, 77
Anxiety. See Psychiatric genetic counseling	tumor gene sequencing
APOE, 49–50, 294	germline implications
APP. See Alzheimer's disease	allelic fraction, 102
Arrhythmogenic right ventricular cardiomyopathy	discordant tumor types, 103
(ARVC), 145, 148	filtration, 101
	founder variants, 102–103
ARVC. See Arrhythmogenic right ventricular	Lynch syndrome case example, 104–105
cardiomyopathy	overview, 101
Assisted reproductive technologies. See Gamete donation;	transcript use, 101–102
Intracytoplasmic sperm injection; In vitro	tumor heterogeneity, 102
fertilization; Preimplantation genetic testing	tumor mutational burden, 103
Attention deficit-hyperactivity disorder.	variant classification, 103–104
See Neurodevelopmental disorders	techniques, 100–101
Autism. See Neurodevelopmental disorders	Cardiovascular disease. See also Sudden cardiac death
В	genetic counseling BATHE assessment, 162–164
BATHE assessment, 162–164	coping promotion, 161–162
Bipolar disorder. See Psychiatric genetic counseling	diagnosis assimilation, 142–143
BRAF, 99	genetic testing
BRCA1, 78, 83, 97, 101–102, 106	decision-making
BRCA2, 78, 83, 101–102, 106	literature, 158–159
Breast cancer, genetic testing, 77–78, 83	overview, 156, 158
	indications, 156–157
C	interpretation, 160–161
-	psychological and social issues
CADD Score. See Combined Annotation	implantable cardioverter defibrillator
Dependent Depletion Score	decision-making, 147
Cancer	outcomes, 147–148
cell-free DNA, 105–106	research findings, 143–146, 159–160
germline variant identification through somatic	sports restrictions, 148–149
analysis, 106–110	sudden cardiac death
Cancer. See also specific cancers	grief, 149–151

Cardiovascular disease. (Continued)	minors, 221
risks, 146-147	personalized medicine, 294
syndromes, 141–142	Down syndrome, preimplantation genetic
CDH1, 88, 109	testing, 202–204, 255
Cell-free DNA (cfDNA)	DTC testing. See Direct-to-consumer testing
prenatal screening	Duty to warn, at-risk relatives, 231–235
aneuploidy, 27–29	•
monogenetic disorders, 29-30	
overview, 15	E
prospects, 34–36	Eating disorders. See Psychiatric genetic counseling
recommendations, 33	ECS. See Expanded carrier screening
tumors, 105–106	EGFR, 103
Cerebral palsy. See Neurodevelopmental disorders	Elective genetic testing
cfDNA. See Cell-free DNA	minors
CHARGE syndrome, 170	direct-to-consumer testing, 221
CHIP. See Clonal hematopoiesis of indeterminate	exceptions to deferment, 217–218
potential	exome/genome sequencing, 218–220
•	prospects, 222
CHMP2B, 51	rationale for discouragement, 214–215
Chromosomal microarray analysis (CMA), 16, 31,	position statements, 213–214
61, 63, 170, 188	*
Clonal hematopoiesis of indeterminate potential	prenatal testing
(CHIP), 105	exome/genome sequencing, 220–221
CMA. See Chromosomal microarray analysis	overview, 216–217
Combined Annotation Dependent Depletion (CADD)	right not to know, 215–216
Score, 173	value assessment, 218
Counsyl Complete, 7	EPCAM, 98–99
Cystic fibrosis	ES. See Exome sequencing
carrier screening, 292–293	Eugenics, 14, 250
genetic counseling, 187, 194	Everolimus, 69
newborn screening, 190	Exome sequencing (ES), 33, 35–36, 170, 218–221
prenatal diagnosis, 32	Expanded carrier screening (ECS), 30, 293
treatment, 192	
	_
D	F
	False-negative rate, cell-free DNA screening, 28–29
DCM. See Dilated cardiomyopathy	False-positive rate, cell-free DNA screening, 28
Decision-making, prenatal genetic testing	Family-centered counseling, pediatric common
challenges, 14–15	diseases, 187-188
educational video, 11–12	Fetal cell, identification in maternal blood, 36
historical perspective, 14	Fetal fraction (FF), cell-free DNA screening, 28-29
nondirective counseling, 12	FF. See Fetal fraction
patient coaching with decision aid, 20-21	FISH. See Fluorescence in situ hybridization
shared decision-making model	FLCN, 97
decision aids, 17-21	Fluorescence in situ hybridization (FISH), 31, 203
overview, 13-14	FMR1, 61
uncertain information and informed decisions, 16	Fragile X syndrome, 63
Depression. See Psychiatric genetic counseling	Frontotemporal dementia (FTD), genetic testing
DGC. See Diffuse gastric cancer	
Diagnostic workup	and counseling, 51–52 FTD. <i>See</i> Frontotemporal dementia
confirmation and results disclosure, 191–192	F1D. See Frontotemporal dementia
test selection and interpretation for pediatrics,	
188–189	G
uncertainty, 189	
•	Gamete donation
Diffuse gastric cancer (DGC), genetic testing, 75, 108–109	donors
Dilated cardiomyopathy (DCM), 143, 145, 158	screening, 208–209
Direct-to-consumer (DTC) testing	types, 209–210
Alzheimer's disease, 49–50	genetic issues, 210
cancer, 78	overview, 207–208
legal issues, 230	Gastric cancer. See Diffuse gastric cancer

<i>GBA</i> , 52–53	Intellectual disability. See Neurodevelopmental disorders
GDD. See Global developmental delay	Intracytoplasmic sperm injection (ICSI), 200
Genetic counseling, definition, 238–239	IVF. See In vitro fertilization
Genetic Information Assistant (GIA), 7	
Genetic Information Nondiscrimination Act	_
(GINA), 226-231, 234, 246	J
Genetic testing, genetic counseling relationship, 5	Jar model, psychiatric disorders, 120-122
GIA. See Genetic Information Assistant	
GINA. See Genetic Information Nondiscrimination Act	
Global developmental delay (GDD).	K
See Neurodevelopmental disorders	Kabuki syndrome, 69, 170
-	KRAS, 99
Н	
HCM. See Hypertrophic cardiomyopathy	L
HD. See Huntington's disease	Laboratory genetic counselor
Health Insurance Portability and Privacy Act	conflicts of interest, 270–271
(HIPAA), 229, 232–235	overview, 263–264
Hereditary nonpolyposis colorectal cancer syndrome.	prospective roles
See Lynch syndrome	business development, 266–267
HIPAA. See Health Insurance Portability and Privacy Act	education of clients and staff, 267–269
Humanism	leadership and management, 267
genetic counseling	research, 271–272
aligning with humanistic goals, 244–246	variant interpretation, 272–273
nonhumanistic aspects, 241–243	roles
role, 239–241	case management, 265
overview, 237–238	communication with clients, 264
Huntington's disease (HD)	result reporting, 264–265
gene discovery, 43–44	test development, 265–266
genetic counseling for predictive testing	work settings, 269–270
prospects, 47–48	Learning disorders. See Neurodevelopmental disorders
protocol, 44–45	Legal issues, genetic testing, 225–235
session characteristics, 46–47	LMNA, 156
predictive testing	Long QT syndrome (LQTS), 143, 146–147, 149, 160
outcomes, 45–46	LQTS. See Long QT syndrome
protocol, 44–45	LRKK2, 52–53
Hypertrophic cardiomyopathy (HCM), 143, 145,	Lynch syndrome
148–149, 158	clonal hematopoiesis of indeterminate potential, 105
140 149, 130	immunohistochemistry, 98–99
	microsatellite instability, 98
I	mismatch repair screening, 99
ICD. See Implantable cardioverter defibrillator	tumor gene sequencing
ICSI. See Intracytoplasmic sperm injection	germline implications
Implantable cardioverter defibrillator (ICD)	allelic fraction, 102
decision-making, 147	case example, 104–105
outcomes, 147–148	discordant tumor types, 103
In vitro fertilization (IVF). See also Preimplantation	filtration, 101
genetic testing	founder variants, 102–103
historical perspective, 199	overview, 101
principles, 200–201	transcript use, 101–102
Informed consent	tumor heterogeneity, 102
alternative models, 281–283	tumor mutational burden, 103
challenges in genomics era, 279–280	variant classification, 103–104
genetic counseling role, 283–284	techniques, 100–101
genomic sequencing impact on models, 278–279	teemiques, 100 101
historical perspective, 276–278	
incidental and secondary findings impact, 280–281	M
prospects, 283–284	MAPT, 51–52
secondary findings, 281	Massively parallel sequencing (MPS), 35

MECP2, 62	Polygenic risk score (PRS)
Million Veterans Program (MVP), 291	Alzheimer's disease, 50
MLH1, 98-99, 104	cancer, 87
Molloy v. Meier, 232	Polygenic risk score, psychiatric disorders, 132-133, 136
MPS. See Massively parallel sequencing	Population genomic sequencing, initiatives, 290-294
MSH2, 98-99, 104	Practice models, genetic counseling, 3-4
MSH6, 98-99, 104	Precision Medicine Initiative, 291
MVP. See Million Veterans Program	Preimplantation genetic testing (PGT)
MyCode, 291–292	aneuploidy, 202–204
	common pediatric diseases, 193
	historical perspective, 250
N	HLA matching, 206–207, 249
NBS. See Newborn screening	mitochondrial disease, 207, 258
NDD. See Neurodevelopmental disorders	monogenetic disease, 205
Neurodevelopmental disorders (NDD)	nondisclosing monogenetic testing, 205–206
definitions, 60	principles, 201–202
etiology, 60-61	
genetic counseling	regulation
decision-making about testing, 65–66	genetic counseling regulation, 255–256, 259
delivery of diagnosis, 67	medical necessity requirement, 256–257
hope fostering, 68	permissiveness degree, 252–253
meaning-making, 68–69	policy approach, 251–252
parental attitudes toward diagnosis, 63–64	reproductive tourism, 258
recurrence risk, 66–67	sex selection, 257–258
•	trends, 254
relationship between genetics and disorders, 64–65	sex selection, 250
social support opportunities, 69	structural chromosome rearrangement, 204-205
stigma, shame, and guilt, 69	variants of uncertain significance, 207
uncertainty management, 67–68	Prenatal genetic testing. See also specific diseases
genetic testing, 61–63	counseling session components, 26-27
treatment prospects, 69–70	decision-making. See Decision-making,
Newborn screening (NBS), referrals, 189-191	prenatal genetic testing
Next-generation sequencing (NGS), 31–32, 62–63, 98, 100	diagnostic tests
NGS. See Next-generation sequencing	aneuploidy, 31
NOTCH1, 110	microdeletions/microduplications, 31–32
	monogenetic disorders, 32
0	recommendations, 32–33
	techniques, 31
OPDG. See Ottawa Personal Decision Guide	patient perspectives, 36
Ottawa Personal Decision Guide (OPDG), 17–19	prenatal treatment options, 36–37
Ovarian cancer, genetic testing, 75, 77–78, 83	
	prospects, 34–36
Р	screening tests
	aneuploidy, 27
Pancreatic cancer, genetic testing, 75–77	cell-free DNA, 27–30
Parkinson's disease (PD), genetic testing and	microdeletions, 29
counseling, 52–53	monogenetic disorders, 29–30
Pate v. Threlkel, 231–232	multiple marker screening, 27
PD. See Parkinson's disease	recommendations, 32
Personalized medicine	reproductive carrier screening, 30–31
genetic counseling as keystone, 296-297	PRS. See Polygenic risk score
overview, 289-290	PSEN1. See Alzheimer's disease
precision health, 290	PSEN2. See Alzheimer's disease
public perspective, 294–296	Psychiatric genetic counseling
PGRN, 51	appointment and follow-up, 118
PGT. See Preimplantation genetic testing	case load, 118
Pheochromocytoma, genetic testing, 76	diagnosis confirmation, 117–118
PMS2, 98–99, 104	historical perspective, 116–117
POLD, 105–106	outcome evaluation, 118, 125–126
POLE, 105–106	overview, 115–116
1 OLL, 10J=100	Over view, 113-110

preappointment procedures, 117	indications, 171–172
process and content	limitations, 171
etiology, 120–122	neonates, 172
familial recurrence chances, 123–124	techniques, 170
initial contracting, 120	variant classification, 172–175
overview, 118–119	neonatal intensive care unit genetic testing and
recovery, 122–123	counseling, 172, 179
session closing, 124	Reciprocal engagement model (REM), tenets for genetic
symptom impact on session, 124–125	counseling, 2–3
prospects, 126	RECQL1, 110
rationale, 116	REM. See Reciprocal engagement model
risk assessment	RET, 83
family history	Retinoblastoma, genetic testing, 76
data collection, 133-134	REVEAL study, 49
provision of risks, 136	
recurrence risks, 123-124, 134-136	
overview, 129-130	S
polygenic risk score, 132-133	Safer v. Estate of Pack, 231-232
prospects, 136-137	Scalability, genetic counseling resources, 5–8
schizophrenia	SCD. See Sudden cardiac death
comorbidity with other disorders, 131	Schizophrenia. See Psychiatric genetic counseling
genetics	SDM model. See Shared decision-making model
copy number variants, 131	Service delivery models, genetic counseling, 5–7
functional categories of genes, 131	Shared decision-making (SDM) model
rare variants, 131	decision aids, 17-21
variants with small effect sizes, 130-131	overview, 13-14
nongenetic risk factors, 132	Siblings, carrier testing, 192–193
team, 117	SIDS. See Sudden infant death syndrome
training, 118	SNCA, 52
PTEN, 62	SOD1, 51
	Sperm donor. See Gamete donation
D.	Sudden cardiac death (SCD)
R	grief, 149–151
Rare disease	psychological impact of young death, 144
definition, 169–170	risks, 146–147
diagnostic odyssey, 177–179	Sudden infant death syndrome (SIDS), 143
genetic counseling, 175	
genetic testing	T
incidental and secondary findings, 175-176	
sequencing	Tarasoff v. Regents of the University of California, 232–233
exome versus genome sequencing, 171	TP53, 88, 101