Subject Index

aCHD, see Adult congenital heart disease
ACTA2, thoracic aortic aneurysm mutations 229, 232, 233
Adult congenital heart disease (aCHD)
acquired cardiovascular disease 76, 77
aortopathy 77
arrhythmias 73, 74
exacerbating factors 71
frequency 32, 70, 71
heart failure 72, 73
pregnancy management 75, 76
psychosocial issues 76
pulmonary hypertension 74, 75
residual, sequelae, and late complications 71, 72
survivors of congenital heart disease and ethics of demand for destination therapy 301, 302
treatment facilities and human resources 71
Alagille syndrome (ALGS)
clinical features
bone 159
cognitive function 162
eye 159, 160
face 160
heart 158, 159
kidney 160, 161
liver 157, 158
overview 156, 157
vasculature 161
diagnosis 161–163
genetic counseling 163
history of study 155, 156
management 162
pathophysiology 156
Alcohol, congenital heart defect risks in pregnancy 66
ALGS, see Alagille syndrome
ALPS, see Autoimmune lymphoproliferative syndrome
Antibiotics, congenital heart defect risks in pregnancy 65
Antidepressants, congenital heart defect risks in pregnancy 54
Antifungals, congenital heart defect risks in pregnancy 65
Aortic aneurysm, see Thoracic aortic aneurysm
Aortic arch
DiGeorge syndrome defects 102
Turner syndrome defects 93–95
Aortic coarctation, see Coarctation of the aorta
Aortic dilation
DiGeorge syndrome 102, 103
Turner syndrome 93
Aortic valvoplasty, interventional cardiac catheterization in neonates 290, 291
Arrhythmia, see also Brugada syndrome;
Catecholaminergic polymorphic ventricular tachycardia; Long QT syndrome; Short QT syndrome adult congenital heart disease 73, 74
prospects for study
Arrhythmogenic ventricular cardiomyopathy (AVC)
clinical features 220
genetics 215, 220, 221
ASD, see Atrial septal defect
Atria
chamber formation 15, 16
septation 19
Atrial septal defect (ASD)
Down syndrome 82, 83, 86
echocardiography 261, 262
historical perspective 6, 7, 9
interventional cardiac catheterization in infants and children 292, 293
monogenetic causes 179–181
Nigeria 47, 52, 53
Turner syndrome 94
Atrial septostomy, interventional cardiac catheterization in neonates 290
Atrioventricular (AV) node, development studies
electrocardiography 18
transcription factors 18
Atrioventricular septal defect (AVSD)
chromosome 8p23.1 deletion 115
CRELD1 mutations 135, 136, 182
Down syndrome 82–87
echocardiography 264, 265
monogenetic causes 181, 182
surgical management in Down syndrome 280–283
Autoimmune lymphoproliferative syndrome (ALPS), gene mutations 136, 137
AV node, see Atrioventricular node
AVC, see Arrhythmogenic ventricular cardiomyopathy
AVSD, see Atrioventricular septal defect
BAV, see Bicuspid aortic valve
Becker muscular dystrophy (BMD), dilated cardiomyopathy association 212, 216
Beta-blockers
catecholaminergic polymorphic ventricular tachycardia management 207
long QT syndrome management 202
thoracic aortic aneurysm management 233
Bicuspid aortic valve (BAV)
monogenetic causes 181
thoracic aortic aneurysm association 229
Turner syndrome 92, 93, 97
Blalock-Taussig shunt 5, 8
BMD, see Becker muscular dystrophy
BrS, see Brugada syndrome
Brugada syndrome (BrS)
clinical manifestations 204
diagnosis 204
epidemiology 204
遗传性变异 204
management 205
risk stratification 205
Cardiac catheterization
historical perspective 4, 8, 9
interventional cardiology, see Interventional cardiac catheterization
Cardiofaciocutaneous syndrome (CFCS), clinical features 125
Cardiomyocyte, see Engineered heart tissue
Cardiomyopathy, see also specific cardiomyopathies
final common pathway hypothesis 211
overview 210, 211
prospects for study 222
Catecholaminergic polymorphic ventricular tachycardia (CPVT)
clinical manifestations 207
diagnosis 206, 208
epidemiology 206
遗传性变异 206
management 207
risk stratification 207
CfLS, see Cornelia de Lange syndrome
CFCS, see Cardiofaciocutaneous syndrome
Channelopathy, see Brugada syndrome;
Catecholaminergic polymorphic ventricular tachycardia; Long QT syndrome; Short QT syndrome
CHARGE syndrome
CHD7
cardiac function 151, 152
genetic defects 145, 146, 148, 149
clinical features 149
clinical surveillance and genetic counseling 152
diagnosis
clinical 146, 147
molecular 147, 148
遗传性-表型关联 149, 150
heart defects 150, 151
overview 145, 146
prospects for study 152, 153
Char syndrome
clinical features 192, 193
diagnosis 193
CHD7
cardiac function 151, 152
CHARGE syndrome defects 145, 146, 148, 149
mutation in syndromes 149
CITED2, germline mutations 136
CNV, see Copy number variation
Coarctation of the aorta
adult congenital heart disease 77
interventional cardiac catheterization stenting in infants and children 294, 295
Cocaine, congenital heart defect risks in pregnancy 66
Computed tomography (CT), atrial septal defect 262
Conotruncal anomaly face syndrome, genetics 101
Copy number variation (CNV)
genetic testing 248–250
Cornelia de Lange syndrome (CdLS)
clinical features 189, 190
diagnosis 190
Coronary arteries, development 21
Cost analysis, see Epidemiology, congenital heart defects
Costello syndrome (CS)
clinical features 125, 126
pathogenesis of cardiovascular disease 128
CPVT, see Catecholaminergic polymorphic ventricular tachycardia
CREBBP, Rubinstein-Taybi syndrome mutations 192
CRELD1
atrioventricular septal defect mutations 182
germline mutations 135–136
heterotaxy defects 171
CRISPR, gene therapy strategy 313, 314
CS, see Costello syndrome
CT, see Computed tomography
DCM, see Dilated cardiomyopathy
Developing countries, see Nigeria, congenital heart defects
Diabetes
congenital heart defect risks in pregnancy 60
maternal diabetes interventions 38, 39
DiGeorge syndrome
cardiac patient
diagnosis 105, 106
management 108
clinical features
congenital heart disease 101
noncardiac manifestations 103, 104
conotruncal defects 104, 105
genetics 101
history of study 100
outcomes 106
surgical management of heart defects 283, 284
Turner syndrome parallels 95, 96
Dilated cardiomyopathy (DCM)
clinical features 212
epidemiology 211, 212
genetics 212–216
muscular dystrophy association 212, 216
DMD, see Duchenne muscular dystrophy
Down syndrome (DS)
atrial septal defect 82, 83, 86
atrioventricular septal defect 82–87
cardiac phenotype 85–87
clinical presentation 85, 87
embryogenesis 84, 85
epidemiology 82, 83
gene therapy prospects 88
genetics 83, 84
surgical management of heart defects 280–284
treatment and prognosis of congenital heart defects 87, 88
ventricular septal defect 82, 83, 85
DS, see Down syndrome
DSG2, arrhythmogenic ventricular cardiomyopathy mutations 221
DSP, arrhythmogenic ventricular cardiomyopathy mutations 221
Duchenne muscular dystrophy (DMD), dilated cardiomyopathy association 212, 216
ECG, see Electrocardiography
Echocardiography, see also Fetal echocardiography
atrial septal defect 261, 262
atrioventricular septal defect 264, 265
Doppler imaging 258
examination overview 259, 260
historical perspective 9
indications 260
physics 257, 258
pulmonary valve stenosis 266, 267
sonographers 260
tetralogy of Fallot 265, 266
timing 260
ventricular septal defect 262–264
EDS, see Ehlers-Danlos syndrome
Edwards syndrome, surgical management of heart defects 284, 285
Ehlers-Danlos syndrome (EDS), thoracic aortic aneurysm association 229
EHMT1, Kleefstra syndrome defects 115, 116
EHT, see Engineered heart tissue
Elastin
supravalvular aortic stenosis defects 181
Williams-Beuren syndrome defects 115
Electrocardiography (ECG), heart development studies
atrioventricular node 18
earliest recordings 18
peripheral ventricular conduction system 18, 19
sinus node 18
Ellis-van Creveld syndrome (EVC)
clinical features 188, 189
diagnosis 189
genetic testing 252
Engineered heart tissue (EHT)
bioreactors 309
cardiomyocyte reentry into cell cycle 312, 313
cell populations 307, 308
challenges
cardiomyocyte maturation 309
immune response 310
ischemia 310
hypertrophic cardiomyopathy applications 313
mechanical load and electrical stimulation 308
prospects 314
scaffolds
deellularized heart 310, 311
extracellular matrix components 307
stem cells 311, 312
EP300, Rubinstein-Taybi syndrome mutations 192
Epidemiology, congenital heart defects, see also specific diseases
adult disease frequency 32, 70, 71
cost analysis
cross-sectional costs 33
lifetime costs 34
overview 32, 33
geographic distribution 31
mortality 34
prevalence
birth prevalence 29, 30
general population 32
variations in reported prevalence 30, 32
prevention
disease types 35, 36
estimation of number of preventable cases 36
folic acid interventions 39, 40
impact maximization 37, 38
initiation 35
integration of interventions 35
maternal diabetes interventions 38, 39
overview 34
preconception care 40, 41
prospects for study 41–43
risk factors, see Risk factors, congenital heart defects
Epilepsy, congenital heart defect risks in pregnancy 64, 65
Ethics
palliative care 302, 303
prenatal diagnosis 299, 300
social justice and congenital heart disease risk 299
survivors of congenital heart disease and demand for destination therapy 301, 302
treatment of children with trisomies 284, 300, 301
EVC, see Ellis-van Creveld syndrome
Fetal echocardiography
examination overview 270–272
importance of fetal diagnosis and outcomes 275, 276
indications 269, 270
interventions for congenital heart defects 271, 273, 274
timing 270
Fever, congenital heart defect risks in pregnancy 64
Folic acid, heart defect interventions 39, 40
Fontan procedure 8
Foramen, septation 19–21
GATA4
atrial septal defect mutations 179, 180
atrioventricular septal defect mutations 182
defects in congenital cardiovascular malformations 115
germline mutations 135
ventricular septal defect mutations 181
Gene reporter assays, clinical significance of transcription factor mutations
chromatin environment 140
isogenic nature 139, 140
mutant protein assays
artificial design 11
cofactors 141
permeability 141, 142
transactivation motifs 141
overview 138, 139
reporter constructs 140, 141
rheostatable promoters 141
Gene therapy
CRISPR strategy 313, 314
Down syndrome prospects 88
targets 313
Genetic testing
Alagille syndrome 163
algorithm 240, 241, 247, 248
CHARGE syndrome 147, 148
copy number variation 248–250
genetic counseling 254
importance of fetal diagnosis and outcomes 275, 276
karyotype testing 248
monogenic diseases 182
overview 238–240
prenatal diagnosis 271
table of diseases, genes and cardiovascular malformations 242–247
targeted gene sequencing 250–252
thoracic aortic aneurysm 229, 230
whole exome sequencing 252, 253
Genome-wide association studies (GWAS), congenital heart defects 182, 239
Genomic rearrangement, congenital cardiovascular malformations 114, 117
GLI2, heterotaxy defects 171
Goldenhar syndrome
clinical features 193
diagnosis 193
GWAS, see Genome-wide association studies

HAND, development role 132, 133
HCM, see Hypertrophic cardiomyopathy
HDAC8, Cornelia de Lange syndrome mutations 190
Health Impact Pyramid 37, 38
Heart development
  chamber formation
    atria 15, 16
    ballooning model 15, 16
    ventricles 17, 18
electrocardiography
  atrioventricular node 18
  earliest recordings 18
  peripheral ventricular conduction system 18, 19
  sinus node 18
  heart tube
    formation 11–14
    growth 12, 15
    insulating plane 21
  prospects for study 22, 23
  septation
    atria 19
    atrioventricular canal and primary foramen 19–21
    outflow tract separation 21
    ventricles 19
  stages 13
  valves
    atrioventricular valves 22, 23
    remodeling in embryo 22
    semiluminar valves 22
    unidirectional flow in primitive heart 22
  vasculature 21
Heart failure, adult congenital heart disease 72, 73
Heart tube
  formation 11–14
  growth 12, 15
Heterotaxy
  ciliopathy association 171, 172
classification 168, 169
complex congenital heart disease 169
diagnosis 167, 168
  genetics 171
left-right patterning and regulation of cardiac looping 169–171
overview 166, 167
prospects for study 173–175
respiratory symptoms 172, 173
Historical perspective, congenital cardiovascular anomalies
  cardiac operations 6–8
correlating anatomic and clinical features 3, 4
diagnostics 4
  early descriptions 3
echocardiography 9
  extracardiac operations 4–6
  interventional catheterization 8, 9
  overview 2, 3
  prostaglandin E therapy 9
HLHS, see Hypoplastic left heart syndrome
Holt-Oram syndrome (HOS), developmental defects 132
HOS, see Holt-Oram syndrome
Hypercholesterolemia, congenital heart defect risks in pregnancy 64
Hyperhomocysteinemia, congenital heart defect risks in pregnancy 64
Hypertension, congenital heart defect risks in pregnancy 64–66
Hypertrophic cardiomyopathy (HCM)
  clinical features 216
  Costello syndrome 125, 126
  engineered heart tissue applications 313
  genetics 216
  genotype-phenotype correlation 217, 218
  Noonan syndrome 124, 125
  Noonan syndrome with multiple lentigines 125
Hypoplastic left heart syndrome (HLHS)
  fetal intervention 273–275, 295
  historical perspective 8
  palliative care
    ethics 302, 303
    hybrid palliation 296
ICD, see Implantable cardioverter defibrillator
Implantable cardioverter defibrillator (ICD)
  Brugada syndrome management 205
  long QT syndrome management 202
  short QT syndrome management 203
Insulating plane, development 21
Interventional cardiac catheterization
  historical perspective 8, 9
  hybrid approaches
    hypoplastic left heart syndrome hybrid palliation 296
    muscular ventricular septal defect closure 296
    overview 295
    paravalvular leak occlusion 297
    hypoplastic left heart syndrome fetal intervention 273–275, 295
infants and children
  atrial septal defect closure 292, 293
  coarctation of the aorta stent implantation 294, 295
Subject Index

pulmonary valve implantation 294
ventricular septal defect closure 293
neonates
aortic valvoplasty 290, 291
atrial septostomy 290
overview 289, 290
pulmonary valvoplasty 291
stent technology 295
Isoproterenol, Brugada syndrome management 205
Jacobsen syndrome, gene mutations 117
JAG1
Alagille syndrome mutations 156
genetic testing 253
JUP, arrhythmogenic ventricular cardiomyopathy mutations 221
Kabuki syndrome
clinical features 187, 188
diagnosis 188
Karyotyping, see Genetic testing
KCNJ2, genetic testing 253
Kleefstra syndrome, EHMT1 defects 115, 116
Klinefelter syndrome, surgical management of heart defects 286
KMD6A, Kabuki syndrome mutations 188
KMT2D, Kabuki syndrome mutations 188
Koolen-de Vries syndrome, gene mutations 116
LDS, see Loeys-Dietz syndrome
Left-ventricular noncompaction cardiomyopathy (LVNC)
clinical features 218, 219
genes 215, 219, 220
LEFTY2, heterotaxy defects 171
Loeys-Dietz syndrome (LDS)
diagnosis 231
gene mutations 227
Long QT syndrome (LQTS)
clinical manifestations 201
diagnosis 199, 201
epidemiology 199
genetic variants 199, 200
management 202
risk stratification 201, 202
LQTS, see Long QT syndrome
LVNC, see Left-ventricular noncompaction cardiomyopathy
Magnetic resonance imaging (MRI)
atrival septal defect 262
tetralogy of Fallot 266
Marfan syndrome (MFS)
diagnosis 230, 231
gene mutations 227
management 233
surgical management of heart defects 285
MCS, see Mechanical circulatory support
Mechanical circulatory support (MCS), surgical management of heart defects 286, 287
MFS, see Marfan syndrome
Monosomy X, see Turner syndrome
Mowat-Wilson syndrome
clinical features 190
diagnosis 190
MRI, see Magnetic resonance imaging
MYBPC3, hypertrophic cardiomyopathy mutations 216, 217
MYH11, thoracic aortic aneurysm mutations 228, 229, 233
MYH6, germline mutations 136
MYH7, hypertrophic cardiomyopathy mutations 216, 217
Myocardium
primary myocardium 15
types 15
Neurofibromatosis type 1 (NF1), thoracic aortic aneurysm association 229
NF1, see Neurofibromatosis type 1
Nigeria, congenital heart defects
age at diagnosis 50, 51
challenges and opportunities 54
clinical presentation 51, 52
etiology 49, 50
management 52, 53
outcomes 54
overview 47
types 47, 48
NIPBL, Cornelia de Lange syndrome mutations 190
NKX2.5
atrial septal defect mutations 179
atrioventricular septal defect mutations 182
development role 132, 133
genereporter assays of mutations 138, 139, 141
 genetic testing 251, 253
germline mutations 134, 135
heterotaxy defects 171
somatic mutations 137, 138
ventricular septal defect mutations 181
Nodal
heterotaxy defects 171
left-right patterning and regulation of cardiac looping 169
Nonsteroidal anti-inflammatory drugs (NSAIDs), congenital heart defect risks in pregnancy 66
Noonan syndrome (NS)
   clinical features 122, 123
   pathogenesis of cardiovascular disease 127, 128
   RAS/MAPK pathway defects and cardiovascular effects 123–125
   thoracic aortic aneurysm association 229
Noonan syndrome with loose anagen hair (NSLAH), clinical features 126
Noonan syndrome with multiple lentigines (NSML)
   clinical features 125
   pathogenesis of cardiovascular disease 128
NOTCH1
   atrioventricular septal defect mutations 182
   bicuspid aortic valve defects 181
   genetic testing 253
   germline mutations 136
NOTCH2, Alagille syndrome mutations 156
NS, see Noonan syndrome
NSAIDs, see Nonsteroidal anti-inflammatory drugs
NSLAH, see Noonan syndrome with loose anagen hair
NSML, see Noonan syndrome with multiple lentigines
Obesity, congenital heart defect risks in pregnancy 64
OFT, see Outflow tract
Outflow tract (OFT), separation 21
Palliative care, ethics 302, 303
Patau syndrome, surgical management of heart defects 284, 285
Patent ductus arteriosus (PDA)
   Char syndrome 192, 193
   historical perspective 2, 4, 5
   Nigeria 47, 52
PCD, see Primary cilia dyskinesia
PDA, see Patent ductus arteriosus
Phenylketonurea (PKU), congenital heart defect risks in pregnancy 65
Pitx2
   atria development role 15
   left-right patterning and regulation of cardiac looping 170, 171
PKP2, arrhythmogenic ventricular cardiomyopathy mutations 221
PKU, see Phenylketonurea
PRDM16, defects in congenital cardiovascular malformations 115
Pregnancy, see Diabetes; Fetal echocardiography;
   Prenatal diagnosis; Risk factors, congenital heart defects
Prenatal diagnosis
   ethics 299, 300
   genetic testing 271
   importance of fetal diagnosis and outcomes 275, 276
Prevention, see Epidemiology, congenital heart defects
Primary cilia dyskinesia (PCD)
   gene mutations 171
   heteroxasy association 172
   respiratory symptoms 172, 173
Prostaglandin E, historical perspective of therapy 9
Pulmonary hypertension, adult congenital heart disease 74, 75
Pulmonary stenosis
   echocardiography 266, 267
   historical perspective 3
Pulmonary valve
   interventional cardiac catheterization implantation in infants and children 294
   valvoplasty via interventional cardiac catheterization in neonates 291
Quinidine
   Brugada syndrome management 205
   short QT syndrome management 204
RAD21, Cornelia de Lange syndrome mutations 190
RASopathy, see also specific diseases
   genetics 126, 127
   genotype-phenotype associations 127
   pathogenesis of cardiovascular disease
      Costello syndrome 128
      Noonan syndrome 127, 128
      Noonan syndrome with multiple lentigines 128
RCM, see Restrictive cardiomyopathy
RERE, defects in congenital cardiovascular malformations 115
Restrictive cardiomyopathy (RCM)
   clinical features 218
   genetics 215, 218
Risk factors, congenital heart defects
   alcohol 66
   antibiotics 65
   antidepressants 54
   antifungals 65
   challenges for study 59
   cocaine 66
   diabetes 60
   epilepsy 64, 65
   fever 64
   hypercholesterolemia 64
hyperhomocysteinemia 64
hypertension 64–66
maternal sociodemographic characteristics
    age 60
    reproductive history 60
    stress 60
measurements of associations 58, 59, 61–63
nonsteroidal anti-inflammatory drugs 66
obesity 64
overview of maternal factors 57, 58
phenylketonurea 65
prospects for study 66, 67
rubella 65
smoking 66
RTS, see Rubinstein-Taybi syndrome
Rubella, congenital heart defect risks in pregnancy 65
Rubinstein-Taybi syndrome (RTS)
    clinical features 191, 192
    diagnosis 192
SGS, see Shprintzen-Goldberg syndrome
Short QT syndrome (SQTS)
    clinical manifestations 203
    diagnosis 203
    epidemiology 202
    genetic variants 202, 203
    management 203, 204
    risk stratification 203
Shprintzen-Goldberg syndrome (SGS), gene mutations 227, 228
Shunts, historical perspective 5
SLOS, see Smith-Lemli-Opitz syndrome
SMC1A, Cornelia de Lange syndrome mutations 190
SMC3, Cornelia de Lange syndrome mutations 190
Smith-Lemli-Opitz syndrome (SLOS)
    clinical features 190, 191
    diagnosis 191
    treatment 191
Smith-Magenis syndrome (SMS), gene mutations 116
Smoking, congenital heart defect risks in pregnancy 66
SQTS, see Short QT syndrome
Statins, Smith-Lemli-Opitz syndrome management 191
Stents, see Interventional cardiac catheterization
Supravalvular aortic stenosis, elastin defects 181
TBX5
    germline mutations 135
    mutation and congenital heart disease 131
TBX20
    atrial septal defect mutations 181
    germline mutations 135
    interaction with other transcription factors 134
    ventricular septal defect mutations 181
Tetralogy of Fallot (ToF)
    DiGeorge syndrome 102, 107
    echocardiography 265, 266
    historical perspective 3, 7
    Nigeria 52, 53
    surgical management 286
TFAP2B, Char syndrome mutations 193
TGFB2, thoracic aortic aneurysm mutations 227
Thoracic aortic aneurysm
    abdominal aortic aneurysm comparison 226
    clinical features 226, 227, 230
    diagnosis 230–232
    gene discovery 227–229
    genetic counseling 229, 230
    management 232, 233
    prospects for study 233
Tissue engineering, see Engineered heart tissue
ToF, see Tetralogy of Fallot
Trisomy 13, see Patau syndrome
Trisomy 18, see Edwards syndrome
Trisomy 21, see Down syndrome
TS, see Turner syndrome
Turner syndrome (TS)
    aortic arch defects 93–95
    aortic dilation 93
    atrial septal defect 94
    bicuspid aortic valve 92, 93, 97
    epidemiology 91
    etiology of cardiovascular defects
        DiGeorge syndrome parallels 95, 96
        fetal lymphedema 94, 95
        neural crest 95
        genetics 96, 97
        genomic imprinting 97
        karyotypes 91, 92
    surgical management of heart defects 28, 286
    thoracic aortic aneurysm association 229
Ultrasound, see Echocardiography
VACTERL association
    clinical features 186, 187
    diagnosis 187
<table>
<thead>
<tr>
<th>Subject Index</th>
</tr>
</thead>
<tbody>
<tr>
<td>Valve development</td>
</tr>
<tr>
<td>atrioventricular valves 22, 23</td>
</tr>
<tr>
<td>remodeling in embryo 22</td>
</tr>
<tr>
<td>semiluminar valves 22</td>
</tr>
<tr>
<td>unidirectional flow in primitive heart 22</td>
</tr>
<tr>
<td>Velocardiofacial syndrome, genetics 100, 101</td>
</tr>
<tr>
<td>Ventricles</td>
</tr>
<tr>
<td>chamber formation 17, 18</td>
</tr>
<tr>
<td>septation 19</td>
</tr>
<tr>
<td>Ventricular septal defect (VSD)</td>
</tr>
<tr>
<td>Down syndrome 82, 83, 85</td>
</tr>
<tr>
<td>echocardiography 262–264</td>
</tr>
<tr>
<td>historical perspective 3, 4, 6, 7</td>
</tr>
<tr>
<td>interventional cardiac catheterization infants and children 293</td>
</tr>
<tr>
<td>muscular ventricular septal defect closure with hybrid technique 296</td>
</tr>
<tr>
<td>monogenetic causes 181</td>
</tr>
<tr>
<td>Nigeria 47, 52, 53</td>
</tr>
<tr>
<td>VSD, see Ventricular septal defect</td>
</tr>
<tr>
<td>WHS, see Wolf-Hirschhorn syndrome</td>
</tr>
<tr>
<td>Williams-Beuren syndrome, elastin defects 115</td>
</tr>
<tr>
<td>Wolf-Hirschhorn syndrome (WHS), gene mutations 116, 117</td>
</tr>
<tr>
<td>ZEB2, Mowat-Wilson syndrome mutations 190</td>
</tr>
<tr>
<td>ZIC3 germline mutations 135</td>
</tr>
<tr>
<td>VACTERL association defects 187</td>
</tr>
</tbody>
</table>