Index

Note: page numbers in italics refer to figures and tables

absent pulmonary valve syndrome 68
academic achievement 150–1
see also education; learning; mathematic skills
achalasia, upper esophageal sphincter 114, 115
adenoid hypoplasia 191–2
velopharyngeal insufficiency 93
adenoidectomy, velopharyngeal insufficiency 93
adenoids, size 188
adolescents 221–2
psychiatric disorders 136
quantitative MRI 170
self-esteem 222
support 228
affective disorders 139
see also depression
air pressure, reduced intraoral 193–4
airway hyper-responsiveness, persistent with bronchospasm 52
amniocentesis
diagnostic 207–8
FISH 209, 210–12
anal malformations 110
anemia, aplastic/hemolytic 129–30
aneuploidy 213, 214
animal models 35
see also mouse models
antibodies
deficits 131–2
functional defects 128
antidepressants 143–4
anxiety disorder 135–6
adolescents 136, 222
aortic arch
anomalies 47, 52
branching 60
isolated 58, 60, 70–1
position 60
with ventricular septal defect 61
artery remodeling 33–4
cervical 52, 58
double 58, 59
interrupted 54–7, 56, 69–70
additional cardiovascular defects 57
pathogenesis 57
surgical techniques 70
types 56, 57, 70
isolated anomalies 58
cardiovascular abnormalities 58
surgical techniques 70–1
persistent fifth 58
right-sided 7, 52, 58
with aberrant left subclavian vein 59
interrupted aortic arch 56
with ventricular septal defect 62
aortic coarctation 63
aortic cusp
deformity 71
prolapse 71
aortic valve
prolapse 71
regurgitation 71
aortopulmonary septation 63–4
aortopulmonary window 57
appetite, cardiovascular dysfunction 112
articulation 94, 95,
192–5
compensatory 194, 194–5
developmental errors 192
multiple errors 192–3
obligatory errors 193–4
articulation therapy 195
artificial insemination by donor 209–10
ARVCF gene 26
aspiration, silent 113–14
astigmatism 116, 117–18
atypia 4–5
atomoxetine 143–4
atrial septal defect, ostium II type 62
atrioventricular canal 63
atrioventricular concordance 63–4
attention 155–6
attention deficit hyperactivity disorder (ADHD) 143–4
attention deficit with/without hyperactivity disorder 135–6
auditory bombardment 192–3
auditory memory 153–4
autistic spectrum disorder 135–6
autoimmune disease 125–7, 129, 129–30

B cells 128, 129
baby in hospital 221
basiranium, obtuse 93
basis pontis 169
BBB syndrome see Opitz syndrome
behavior
abstract 154–5
outcomes 159
behavior modification techniques 144
behavioral difficulties/disorders
attention problems 155–6
behavior modification techniques 144
children 135–6
chromosome 22q11 deficiency syndrome 35
withdrawn behavior 136, 139
early treatment 141–2
behavioral phenotype 139
environmental factors 159
 genetic factors 159
bipolar disorders 9, 135–6
bland affect with minimal facial expression/ extremes of behavior 135
blood products, irradiated 71–2, 124, 130
CMV-negative 131
blowing exercises 189–90
bolus
esophageal phase 114
formation by chewing 110–11
propagation 113
bone marrow transplantation 127, 131
Bourdon–Vos dot test 155–6
brachiocephalic arteries, anomalies 60
brain
activation patterns with functional MRI 167–8
chromosome 22q11 deletion origin effect on development 173
dysmaturation 172
functional imaging 151–2, 183
brain activation patterns 167–8
left lateral fissure 167–8
volume
chromosome 22q11 deletion origin 173–4
in schizophrenia 172
see also regions of brain
brain abnormalities 118
midline defects 165–6
MRI 165–6
schizophrenia 169–70
volume changes 166–7
branchial arches, anomalies 58
branchio-oto-renal syndrome 106
breastfeeding 115
breathing coordination with swallowing/sucking 113–14
bronchitis, recurrent 125–7
bronchomalacia 52
BRUNOL3 gene 37
bupropion 143–4
cardiac anomalies 2
candidate genes 7
congenital 5–7
feeding difficulties 112
conotruncal 6, 7, 47, 210–12
chromosome 22q11 deficiency syndrome 65
chromosome 22q11 deletion association 48
diagnostic methods 65–6
outflow tract anomalies 48
surgical repair 67
identification 124
intellectual outcome 148
major risk sites 64
mouse models 32
outflow tract 48
cardiac surgery, parental information 225–6
cardiocutaneous syndrome 6
cardiovascular defects 47–9
clinical implications 65–6
common findings 63–5
congenital 212, 212
deletion screening 66
feeding difficulties 112
metabolic requirements 112
mortality 47
neonates 65
perioperative policy 71–2
phenotypes 49
prenatal diagnosis 65
prevalence 48–9
surgical implications 66–72
surgical repair 67
types 48–9, 49–65
carotid artery, internal
anomalies 99
displacement 185–6
medial deviation 100
CATCH 22 syndrome 13, 14
rejection as diagnostic label 15
catechol-O-methyltransferase (COMT), gene
coding for 140
chromosome 22q11 141
CD3 counts 126, 129
CD4 counts 126, 129
CD8 counts 126
CDC45L gene 25
CDCREL1 gene 25
central velar translucency 85–6
cerebellum
cytoarchitecture 169
development 168
structural alteration 168
vermal size 169, 170
volume 170
cerebral arteries 166
cerebrospinal fluid volume in schizophrenia 172
cervical spine anomalies 12
CHARGE association 11
chromosome 22q11 deletion 123
chewing, bolus formation 110–11
chorioretinal coloboma 117
chromosome 10p deletion syndromes 36–8
DiGeorge sequence 107
chromosome 22q11
band rearrangements 7–8
COMT gene 26
coding for 141
mapping to 140
low copy repeats 29–30
neural cell migration/differentiation 141
non-random rearrangements 29
PRODH gene mapping to 140, 141
repeat sequences 29–30
susceptibility genes mapping to 141
chromosome 22q11 deficiency syndrome
behavioral difficulties 35
chromosomai architecture disruption 36
cognitive deficits 35
conotruncal defects 65
immunodeficiency 123–4, 125–7
numerical skills deficit 152
screening
guidelines 66
indication 58
visuospatial deficit 152
chromosome 22q11 deletion 19, 200–2
associated syndromes 12–13, 14–15, 123
chromosome engineering experiments 32
cleft of secondary palate 87
cleft palate association 84–5
concomitant diagnoses 200–2
conotruncal heart defects 48
embryology of structures affected 20
families 203
findings in unselected patients 208
incidence 200, 201, 210–12
inheritance 204, 205
mechanism 29–30, 30
mortality 202
mosaicism 29, 206
origin 159
brain development 173
brain volume 174
language development 157
parent of origin 202–4
parental 206, 208–9, 210
inversion polymorphisms 30
prenatal diagnosis 19–20
recurrence risk 204–6, 205, 207, 211
parental knowledge 208–9
schizophrenia 8–9, 139
size 159
tetralogy of Fallot 214
variability 202
velopharyngeal dysfunction 91
DiGeorge sequence 5, 10–11
causes 107
chromosome 22q11 deletion 123
immunodeficiency 125–7
VCFS association 7–8
DiGeorge syndrome 14
chromosome deletions 14–15
mortality 202
Down syndrome 183
ductus arteriosus
absence 50
anomalous 64
left pulmonary artery arising from 53–4
patent 63
left-sided 52
dysmorphisms 2
eating 222–3
see also feeding difficulties
echocardiography, fetal 210–12
education
academic achievement 150–1
family issues 223–5
homework 225
individual plans 224
organizational skills 225
provision 144
special needs 227
see also learning; mathematic skills
embryology, structures affected in 22q11DS 20
embryonic developmental abnormalities 141
embryonic recovery 32–3
embryotoxon, posterior 116
emotional instability 135–6
endocrine disorders 4–5
enuresis 105–6
nocturna 109
environmental factors, behavioral phenotype 159
epiglottis 113
eponyms 13–14
esophageal atresia 110
esophageal phase of feeding 114–15
normal development 114
VCFS 114–15
esophageal sphincter
lower 114
upper 113, 114
esophagus, peristaltic waves 114
executive function 154–5
mathematic problem-solving skills 153
eye
fundus 117
ophthalmic manifestations 116–18
retinal vascular tortuosity 116, 116–17, 117
suborbital blue coloring 116
see also visual entries
facial dysmorphism 2, 5
feeding difficulties 112
facial musculature, flaccid 3, 4
familial recurrence of VCFS 65, 208–9
family
chromosome 22q11 deletion 203
interaction styles 159
involvement in psychiatric disorders 143
physician help 225–7
support 226
see also parents
family history 214
father, impact of initial diagnosis 220
feeding difficulties 110–11, 124, 136
breastfeeding 115
cleft palate 111–12
developmental influence 115
fetal echocardiography 210–12
fibroblast growth factors (FGFs) 34
fluorescent in situ hybridization (FISH) analysis 130, 214
amniocentesis 209, 210–12
fragile X syndrome 169
frontal lobe
changes in schizophrenia 169–70
enlargement 166–7
fronto-parietal polymicrogyria 166
fronto-temporal connectivity, disrupted 172–3
Furlow technique see Z-plasty technique, double-opposing (Furlow)
G syndrome see Opitz syndrome
gastric emptying 112
gastrointestinal system
congenital malformations 110
hypomobility 112
manifestations 109–16
GATA3 gene 37
haploinsufficiency 107
mutation/deletion in HDR syndrome 123
general practitioners 226, 227
genes
expression modulation in imprinting 173
mapping within TDR regions 21–9
mouse models 31–3
testing candidate 31
genetic counseling 200–2, 206–9
diagnosis 207–8
parental studies 210–12
genetic factors, behavioral phenotype 159
germline mosaicism 19–20, 204
glottal stop 194–5
 glutamatergic synapses 141
GNB1L gene 26
GP1B3 gene 25–6
graft-versus-host disease 125–7
prevention 131
grandparents, impact of initial diagnosis 220–1
gray matter
chromosome 22q11 deletion origin 173–4
deficit 170–1
volume in schizophrenia 171–2
decrease rate 174–5
risk for children 174–5
great arteries, transposition 57, 62
GSCL gene 23
guidelines, prenatal diagnosis 209–14
haploinsufficiency 21, 31
GATA3 gene 107
phenotype production 31
TBX1 gene 21, 36
polymicrogyria relationship 166
HC-II gene 28
health visitors 226, 227
hearing loss, conductive/sensorineural 182–3
heart disease, congenital 212, 212
Hira gene 24
Hirschsprung’s disease 110
hoarseness 185–6, 187
treatment 187
holoprosencephaly sequence 11
humoral immunity assessment 131–2
hydronephrosis 213
hypermastia 116
hypernasality 3, 6, 187–8
compensatory articulation errors 195
diagnosis 190–1
perceptual evaluation 190–1
quantification 190–1
surgical treatment 191
treatment 189–90
hypertension, childhood 108
hypocalcemia, neonatal 65, 124
hyponasal speech 191–2
pharyngeal flap surgery 99–101
hypoparathyroidism 5
nephrocalcinosis 109
hypoparathyroidism, sensorineural deafness and renal anomalies (HDR) syndrome 37, 107
GATA3 mutation/deletion 123
hypoplastic left heart syndrome 63
hypotonia, velopharyngeal musculature 94
immune disorders 4–5
immune system compromise 223
immunodeficiency 123–32
chromosome 22q11 deficiency syndrome 123–4, 125–7
consequences 129–30
correlation lack with phenotypic features 127–8
DiGeorge sequence 125–7
natural history 128
immunoglobulin(s) 129
immunoglobulin A (IgA) 129
deficiency 131–2
immunologic evaluation 128
imprinting 174
infections
fungal 125–7
management 130
morbidity 130
mortality 125–7, 130
recurrent 124, 131–2
respiratory 125–7
secondary bacterial 125–7
influenza vaccination 130
information storage/retrieval, parietal lobe
dysfunction 167
infundibular septum, hypoplastic/absent 50, 63–4
interrupted aortic arch 56–7
surgical technique 67
infundibuloarterial inversion, isolated 63
inheritance 204
intellectual impairment 148
intellectual outcome, heart anomalies 148
intellectual profile 149–50
intelligence 148
crystallized 149
fluid 149
Index

intraoral air pressure 193–4
intraoral examination 95
intravenous immunoglobulin 131–2
in-vitro fertilization (IVF) 209–10
IQ
borderline 167
full-scale 148
performance 149, 150, 151
VIQ discrepancy 157–8
verbal 149, 150, 151
PIQ discrepancy 157–8
iris coloboma 117
jejunal atresia 110
Joubert syndrome 169
kidney
dysplastic/multicystic 213
see also renal entries
language
delay 86–7, 156–7, 181–2
causes 182–3
chromosome 22q11 deletion origin 157
difficulties
non-verbal learning 158
parietal lobe abnormalities 167
disorders 181, 181–4
interventions 183–4
syndrome-related 182–3
expressive 156–7, 181–2, 182
oral approach 183–4
receptive 156–7
sign 183–4
specific impairment 182
use 224
laryngeal complex 113
laryngeal web 185–6, 187
neck 11
resection 186–7
larynx, asymmetry 185–6
lateralization defects 63
learning
characteristics 184
curve 184
environment 124–5
home programs 184
methods 184
non-verbal 182
language difficulties 158
problems 227–8
rote 224
verbal 153–4
see also education
learning disability 117–18, 148
non-verbal 136, 157–8
parietal lobe abnormalities 167
levator palatini muscles, velopharyngeal closure 91–2
levator sling reconstruction 89
levator veli palatini 84–5
dissection and repositioning 88
double-opposing Z-plasty technique 89
submucosal cleft palate diagnosis 85–6
levocardia 63–4
limb anomalies 213
lip
short upper and feeding difficulties 112
see also cleft lip
lymphocytes
counts 130
proliferation responses 130
lymphomas 125–7
LZTR1 gene 29
magnetic resonance angiography (MRA), cerebral arteries 166
magnetic resonance imaging (MRI)
functional 151–2, 183
brain activation patterns 167–8
qualitative studies 166
quantitative
adolescents 170
adults 173
children 170
major aortopulmonary collateral arteries (MAPCA) 52
surgical technique 68–9
malignancy 130
management 130–2
multidisciplinary approach 124–5
mathematic skills 150–1, 151–2, 151–3
difficulties 154
problem-solving 153
processing systems 152
medical issues 223
memory 153–4
auditory 153–4
parietal lobe role 167, 183
rote verbal 154
spatial working 153–4
verbal 154
visuo-spatial 117–18, 154
test 153–4
working 183
metabolic requirements, cardiovascular defects 112
methylphenidate 143–4
microcephaly 167
microdeletion syndromes 20–1
miscarriage rate 209
molecular genetics 19, 20–33
monoaminergic neurotransmission 141
mood swings in adolescents 222, 228
mortality 202
mosaicism 204, 206
chromosome 22q11.2 deletions 29
germline 19–20, 204
mother, impact of initial diagnosis 219
mouse models 31–3
Df1 mice 35
embryonic recovery 32–3
shortest region of deletion overlap (SRDO) mapping 33
mucoperiosteal flaps, cleft palate surgical management 88
musculus uvulae 86
absence/hypoplasia 83–4
velopharyngeal closure 91–2
nasal air emission 94, 94–5, 193–4
nasal turbulence 193–4
nasendoscopy 95–6
flexible fiberoptic 95
nasometer 190–1
nasopharyngoscopy 190–1
natural killer cells 128, 129
neck, webbing 11
need ratio 93
neonates, cardiovascular defects 65
neo-pulmonary artery index, total 69
nephrocalcinosis 105–6, 109
nephro-urological anomalies 105–9
pathogenesis 106–7
screening 108
neural cell migration/differentiation 141
neural crest
cell reduction and persistent truncus arteriosus 54
development 20
abnormalities in VCFS 141
developmental genetics 33–5
neural tube defects 213
neuroimaging 165–76
NLVCF gene 24
nonverbal ability 149–50
Noonan syndrome 11, 12
chromosome 22q11 deletion 123
nosology 13–15
number recall 153–4
numerical skills 151–3
deficits 152
neural disruption vulnerability 152–3
nutrition
impaired 124
optimization in infants 131
object memory test 153–4
obessive behavior 222–3, 224
obstructive sleep apnea 99–101
obturator prosthesis 97
occipital lobe, white matter reduction 167
occupational therapists 227
oculo-auriculo-vertebral dysplasia spectrum (OAVS) 11, 12
olanzepine 142
oligohydramnios 109
ophthalmic manifestations 116–18
Opitz syndrome 12–13
chromosome 22q11 deletion 123
optic discs, small 116
oral cavity examination 95
oral phase of feeding 111–12
normal development 111
preparatory 110–11
VCFS 111–12
organizational skills 225
oropharynx examination 95
ostial stenosis 64
with diffuse hypoplasia 53–4
otitis media, recurrent 125–7
otolaryngologists 94
P2X6 gene 29
palatal lift prosthesis 97
palate
anomalies 83
clefts in secondary 87
congenitally short 3
hard 85–6
lengthening 88, 90
phenotype definition 83–8
palate (cont.)
soft
displacement 92
lowering 111–16
velopharyngeal valve 91
surgical decisions 223
V-Y repositioning 88
palatoglossus muscle contraction 111–16
palatopharyngeal disproportion 93
palatopharyngeus muscles 91–2
absent left 84
palpebral fissures, narrow 116
papillo-renal syndrome 106
parent of origin 202–4
parents affected 206, 208–9, 210
inversion polymorphisms 30
baby in hospital 221
children’s learning 184
with chromosome 22q11 deletion 210
with chromosome 22q11.2 deletion 208–9
with chromosome 22q11.2 deletion 208–9
genetic studies in genetic counseling 210–12
home programs for learning 184
impact of initial diagnosis 219–20
information for 223–5
labeling by health professionals 226
parietal lobe
abnormalities 167, 183
function alteration 167–8
inferior 152
white matter reduction 167
Passavant’s ridge 92
PAX2 gene 106–7
PCQAP gene 28
pediatricians 227
Perceptual Organization 149, 150
performance IQ 149, 150, 151
VIQ discrepancy 157–8
personality 222–3
pharyngeal depth 93, 188–9
pharyngeal flap surgery 90, 191
complications 99–101
hyponasality 191–2
posterior 99
velopharyngeal dysfunction 90–1, 98
pharyngeal hypotonia 189
pharyngeal phase of feeding 112–14
normal development 112–13
VCFs 113–14
pharyngeal structure loss 34
pharyngeal volume 188–9
pharyngeal walls lateral 188–9
medial displacement 92
posterior 190
soft tissue 189
velopharyngeal valve 91
pharyngoplasty
internal carotid artery anomalies 99
sphincter 100
velopharyngeal dysfunction 90–1, 98
phenotype
behavioral 159
cardiac 49
delineation 5
Df(1) deletion 32
diversity 124
haploinsufficiency of different genes 31
investigation 160
overlap 12
palatal definition 83–8
second developmental sequences 8
speech effects 186
variation 19
see also cognitive phenotype
phonation 94, 185
phonological errors 192–3
physicians
education 227
help for individuals/families 225–7
physiotherapists 227
PIK4CA gene 28
plastic surgeons 94
platybasia 93, 188–9
PNUTL1 gene 25
polydactyly 213
polyhydramnios 213
polymicrogyria, fronto-parietal 166
pons
cytarchitectures 169
development 168
posterior fossa, aberrant size 169
Potter sequence 11, 109
pragmatic skills 181–2
pregnancy termination, therapeutic 209
pre-implantation diagnosis 209–10
prenatal diagnosis 210
chromosome 22 q11.2 deletions 19–20
guidelines 209–14
presentation, atypical/delayed 131
problem solving 154–5
proline dehydrogenase (PRODH) gene 21–2, 140
chromosome 22q11 141
prosthetic devices, velopharyngeal insufficiency 97
prosthodontists 97
Psychiatric Assessment Schedule for Adults with Developmental Disability 142
psychiatric disorders
adolescents 143–4
adults 136–8, 142
assessment 141–4
children 135–6, 143–4
diagnostic tools 142
early manifestations 182
eyearly treatment 141–2
family involvement 143
late onset 8
manifestations 8–9
neuropsychological assessment 143
rates 137
treatment 141–4
response in adults 142
psychotic disorders 137–8
adolescents 143
brain dysmaturation 172
eyearly treatment 141–2
pulmonary arteries
absent left 51
anomalies with ventricular septal defect 61
confluent 52
crossing 53–4, 64
discontinuous 50, 53–4, 64
hypoplasia 64
isolated inversion 63
nonconfluent 69–70
unilateral absence 68
pulmonary atresia with ventricular septal defect 50–3
additional cardiovascular defects 52, 53
surgical repair 68–9
pulmonary valve
absence 50, 51
anomalies 64
pyelography 108
RANBP1 gene 27
rating scales 94
reading skills 150–1
recombination 30
recurrence risk 204–6, 205, 207, 211
parental knowledge 208–9
refractive errors 116
correction 117–18
renal agenesis 105–6, 213
bilateral 109
unilateral 106, 108
renal dysplasia, multicystic 105–6
bilateral 109
renal failure, chronic childhood 108
renal ultrasound 108, 213
renography 108
resonance 94, 187–92
disorders 191–2
respiratory syncytial virus prophylaxis 130
respite care 228
retinal vascular tortuosity 116, 116–17, 117
rheumatoid arthritis, juvenile 129–30
right ventricle, double outlet 62
risperidone 142
Robin sequence 10–11
rote learning, verbal 153–4
rote memory, verbal 154
RTN4R gene 27
schizophrenia 8–9, 137–41
brain
abnormalities 169–70
volume 172
chromosome 22q11 deletion 8–9, 139
COMT gene coding for 140
CSF volume 172
frequency in VCFS 138–9
fronto-temporal connectivity disruption 172–3
gray matter volume 171–2
decrease rate 174–5
risk for children 174–5
mechanism in VCFS 141
neurodevelopmental disorder 141
precursor symptom identification 142
premorbid neurodevelopmental etiology 174–5
PRODH gene coding for 140
social problems prior to 169
structural neuroimaging abnormalities 141
susceptibility locus mapping to 22q11 140–1
Tbx1 transcription factor 140–1
VCFS association 138–41
white matter
diffusion tensor imaging 171–2, 175
volume 171–2

© in this web service Cambridge University Press

www.cambridge.org
school issues 124–5, 221
adolescents 221–2
SCL25A1 gene 23
secondary developmental sequences 8, 9–12
selective serotonin reuptake inhibitors (SSRIs) 142, 143
self-esteem in adolescents 222
semilunar valves, dysplastic 64
sensorimotor deficits, parietal lobe abnormalities 167
sequence definition 10
services, centralized 226
shift of set 154–5
shortest region of deletion overlap (SRDO) mapping 31
mouse model 33
siblings, impact of initial diagnosis 220
single gene hypothesis 31
sinus solitus, visceral-atrial 63–4
SLC7A4 gene 29
SNAP29 gene 28
social difficulties 224, 227–8
brain alterations 169
social issues 221–2
social skills, poor 136
social withdrawal 136, 139
brain alterations 169
early treatment 141–2
see also withdrawn behavior
social workers 227, 228
spatial cognition 153
spatial working memory 153–4
speech and language therapists 227
speech pathologists 94
multiple articulation errors 192–3
procedures 187
speech therapy 94
contraindications 193–4
hypernasal speech treatment 189–90
presurgical 95
speech-bulb appliances 191
speech/speech disorders 3–4, 181, 184–5
abnormalities 156, 184–5
cleft palate 193–4
compensatory 194, 194–5
compensatory errors 193–4
delay 156–7
developmental errors 192
expressive 181–2
glottal 194–5
interventions 183–4
outcome after cleft palate surgery 90
patterns 184–5
perceptual assessment 94
phenotype 186
syndrome-related 182–3
velopharyngeal closure 92
velopharyngeal dysfunction 91
see also hypernasality; hyponasal speech
spelling skills 150–1
sphincter pharyngoplasty, velopharyngeal dysfunction 90–1, 98
spina bifida occulta 12
spinal cord, tethered 12
SREC2 gene 28
stereotypic personality 135
strategy formation 153–4
structural anomalies 214
subaortic resection 70
subaortic stenosis 70
subclavian artery
aberrant 52, 58
abnormalities with interrupted aortic arch 56
cephalic 52
suborbital blue coloring 116
sucking, infants 110–11
cardiovascular dysfunction 112
coordination with swallowing/breathing 113–14
non-nutritive 111–16
support groups 223, 226
support services 227–8
adolescent 228
supramarginal gyrus
activation 167–8
mathematical processing 152
supramarginal gyrus, left 151–2
surgery
decisions 223
parental information 225–6
see also cleft palate, surgical management; named techniques
swallowing
coordination with breathing/sucking 113–14
difficulties 110–11
pharyngeal phase 112–13
syndrome
definition 1, 10
early descriptions 1
recognition 2–3
syntax delays 181–2
syrrinx 12

T cells 126
bone marrow transplantation 131
function 125
functional defects 128, 129
number indicator 130
production decline 128, 129
screening for defects 128
thymus transplantation 131
T10 gene 27
Takao syndrome see CATCH 22 syndrome
Tbx1/− embryo, pharyngeal structure loss 34
TBX1 gene 7, 33, 140–1
adjacent gene deletion 36
haploinsufficiency 21, 36
polymicrogyria relationship 166
mutations 33–4
occurrence in nondeletion VCFS 35–6
regulation 35
TCFL2 gene 29
TDR regions 21
candidate genes 31
genes mapping within 21–9
temporal lobe changes in schizophrenia 169–70
teratogens, DiGeorge anomaly 107
termination, therapeutic 209
tetralogy of Fallot 9–10, 49–50, 51
additional cardiac defects 50, 50
chromosome 22q11.2 deletion 214
extracardiac anomalies 49
genetic syndromes 49
incidence 212
infundibular septum hypoplasia/absence 50
intracardiac repair 68
pulmonary artery unilateral absence 68
with pulmonary atresia 51–2
pulmonary valve absence 50
surgical repair 67–8
tetrasomy 12p 213
therapists 227
thrombocytopenic purpura, idiopathic 129–30
thymus
aplasia 202
congenital absence 4–5
hypoplasia 128, 202
transplantation 127, 131
thyroiditis, autoimmune 129–30
tonsillar hypertrophy 95, 191–2

inféro-posterior displacement 185–6
pharyngeal flap surgery 99–101
tonsillectomy 192
tracking 155–6
tricuspid atresia 63
truncus arteriosus
additional cardiovascular defects 54
angiocardiology 55
persistent 53–4
surgical repair 69–70
TRXR2 gene 26
TSK gene 23
TUPLE1 gene 24
TUPLE1 transcription factor 14
Turner syndrome 11, 12
UFD1I gene 7, 24–5
ultrasound
prenatal diagnosis 209, 210–12
renal 108, 213
uncinate fasciculus 172–3
ureteric bud
abnormal development 106–7
vascular disruption 107
urinary system
infections 108
manifestations 106
uropathy 106
bilateral 106
congenital
clinical manifestations 108
frequency 108
prenatal presentation 108–9
vascular disruption 107
USP18 gene 27
uvula, bifid 83–4
submucosal cleft palate
diagnosis 85–6
occult 86
vaccines, live viral avoidance 130, 131–2
variability, inter-/intrafamily 202
varicella 131–2
varicella zoster immunoglobulin (VZIg) 131–2
vascular endothelial growth factors (VEGF)
metabolic modifier 159
vascular malformation, ureteric bud 107
vascular smooth muscle cells, neural crest-derived
33–4
<table>
<thead>
<tr>
<th>Page</th>
<th>Index</th>
</tr>
</thead>
<tbody>
<tr>
<td>88</td>
<td>Veau–Wardill–Kilner technique</td>
</tr>
<tr>
<td>84</td>
<td>velar cleft</td>
</tr>
<tr>
<td>91–2</td>
<td>velar eminence</td>
</tr>
<tr>
<td>90</td>
<td>velar hypotonia</td>
</tr>
<tr>
<td>92</td>
<td>velar stretch</td>
</tr>
<tr>
<td>85–6</td>
<td>velar translucency, central</td>
</tr>
<tr>
<td>91–2</td>
<td>Velo–Cardio–Facial Syndrome Educational Foundation</td>
</tr>
<tr>
<td>188</td>
<td>velopharyngeal closure</td>
</tr>
<tr>
<td>187–8</td>
<td>inadequate</td>
</tr>
<tr>
<td>92</td>
<td>speech</td>
</tr>
<tr>
<td>90–1,98</td>
<td>submucosal cleft palate surgical management</td>
</tr>
<tr>
<td>90</td>
<td>velopharyngeal disproportion</td>
</tr>
<tr>
<td>93</td>
<td>velopharyngeal dysfunction</td>
</tr>
<tr>
<td>95</td>
<td>articulation</td>
</tr>
<tr>
<td>91</td>
<td>communication</td>
</tr>
<tr>
<td>93</td>
<td>etiology</td>
</tr>
<tr>
<td>94–5</td>
<td>feeding difficulties</td>
</tr>
<tr>
<td>94–3</td>
<td>nature in VCFS</td>
</tr>
<tr>
<td>98</td>
<td>pharyngeal flap surgery</td>
</tr>
<tr>
<td>90–1</td>
<td>pharyngoplasty</td>
</tr>
<tr>
<td>90</td>
<td>secondary management</td>
</tr>
<tr>
<td>91</td>
<td>speech</td>
</tr>
<tr>
<td>98</td>
<td>sphincter pharyngoplasty</td>
</tr>
<tr>
<td>86</td>
<td>submucosal cleft palate</td>
</tr>
<tr>
<td>90–1</td>
<td>variable</td>
</tr>
<tr>
<td>94–6</td>
<td>velopharyngeal function assessment</td>
</tr>
<tr>
<td>96</td>
<td>velopharyngeal gap</td>
</tr>
<tr>
<td>91</td>
<td>velopharyngeal insufficiency</td>
</tr>
<tr>
<td>93</td>
<td>adenoid hypoplasia</td>
</tr>
<tr>
<td>97</td>
<td>nonsurgical management</td>
</tr>
<tr>
<td>93</td>
<td>post-adenoidectomy</td>
</tr>
<tr>
<td>98</td>
<td>prevalence</td>
</tr>
<tr>
<td>97</td>
<td>prosthesis devices</td>
</tr>
<tr>
<td>98</td>
<td>severity</td>
</tr>
<tr>
<td>190–1</td>
<td>speech-bulb appliances</td>
</tr>
<tr>
<td>95</td>
<td>surgical management</td>
</tr>
<tr>
<td>95</td>
<td>velopharyngeal mechanism, visualization</td>
</tr>
<tr>
<td>94</td>
<td>velopharyngeal musculature hypotonia</td>
</tr>
<tr>
<td>189–90</td>
<td>velopharyngeal region, superior constrictor muscle</td>
</tr>
<tr>
<td>95</td>
<td>velopharyngeal screening, intraoral examination</td>
</tr>
<tr>
<td>91–2</td>
<td>velopharyngeal valve, anatomy/physiology</td>
</tr>
<tr>
<td>91</td>
<td>velopharyngeal valving</td>
</tr>
<tr>
<td>91–6,188</td>
<td>velopharyngeal insufficiency</td>
</tr>
<tr>
<td>187–8</td>
<td>velopharyngeal disproportion</td>
</tr>
<tr>
<td>93</td>
<td>speech</td>
</tr>
<tr>
<td>91</td>
<td>sphincter pharyngoplasty</td>
</tr>
<tr>
<td>57</td>
<td>ventricular septal defect</td>
</tr>
<tr>
<td>61</td>
<td>cardiovascular abnormalities</td>
</tr>
<tr>
<td>70</td>
<td>closure</td>
</tr>
<tr>
<td>57</td>
<td>double committed</td>
</tr>
<tr>
<td>59–61</td>
<td>subarterial</td>
</tr>
<tr>
<td>59–61</td>
<td>subaortic</td>
</tr>
<tr>
<td>62</td>
<td>surgical techniques</td>
</tr>
<tr>
<td>63–4</td>
<td>subaortic</td>
</tr>
<tr>
<td>71</td>
<td>verbal ability</td>
</tr>
<tr>
<td>149–50</td>
<td>verbal Comprehension</td>
</tr>
<tr>
<td>150</td>
<td>verbal impairment</td>
</tr>
<tr>
<td>151</td>
<td>verbal IQ</td>
</tr>
<tr>
<td>157–8</td>
<td>PIQ discrepancy</td>
</tr>
<tr>
<td>153–4</td>
<td>verbal learning, rote</td>
</tr>
<tr>
<td>154</td>
<td>verbal memory</td>
</tr>
<tr>
<td>151</td>
<td>verbal skills</td>
</tr>
<tr>
<td>105–6</td>
<td>vesico-ureteric reflux</td>
</tr>
<tr>
<td>153</td>
<td>visual object cognition</td>
</tr>
<tr>
<td>150</td>
<td>visuo-spatial abilities</td>
</tr>
<tr>
<td>152</td>
<td>deficits</td>
</tr>
<tr>
<td>151</td>
<td>impairment</td>
</tr>
<tr>
<td>154</td>
<td>test</td>
</tr>
<tr>
<td>136</td>
<td>visuo-perceptual skills</td>
</tr>
<tr>
<td>181–2</td>
<td>vocabulary, delays</td>
</tr>
</tbody>
</table>
vocal folds
  airway protection 113
  atrophy 187
  hyperfunction 187
  incomplete adduction 185–6
  laryngeal web 185–6
  paralysis 187, 187
  unilateral 185–6
  vibration 185
  voice 185–7
  pitch 185, 185–7
  disorders 186–7
  volume 185
  see also hoarseness

Weigl's test 154–5
white matter
  chromosome 22q11 deletion origin 173–4
deficit 170, 170–1
  diffusion tensor imaging 172–3
  schizophrenia 171–2, 175
  hyperintensities 165–6
  reduction in posterior brain 167
  volume 171
  schizophrenia 171–2
  Williams syndrome 152, 169
  Wisconsin Card Sorting test 154–5
  withdrawn behavior 136, 139
  see also social withdrawal

ZNF74 gene 27
Z-plasty technique, double-opposing (Furlow) 89, 89, 90, 90–1