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RETIRED *GeneReviews*

Created: October 10, 2019; Revised: June 6, 2024.

For a variety of reasons, the *GeneReviews* Editors occasionally remove chapters from the active website. See the table for a list of the chapters that have been retired and the reasons why they were retired. An archived version of these chapters may be accessed; see Note following table.

Retired *GeneReview* Chapter Title	Reason	Date Retired	SHORTNAME
15q24 Microdeletion Syndrome	1	5-9-19	mdel15q24
1p36 Deletion Syndrome	1	8-8-19	del1p36
22q11.2 Duplication	12	1-30-20	dupl22q11
2q37 Microdeletion Syndrome	1	1-18-18	del2q37_2
9q22.3 Microdeletion	1	8-2-18	mdel9q22_3
Adams-Oliver Syndrome	4	8-17-23	adams-oliver
APOE p.Leu167del-Related Lipid Disorders	9	11-7-19	apoe-leu167del
Ataxia with Oculomotor Apraxia Type 1	4	5-2-24	aoa
CATSPER-Related Male Infertility	16	12-14-23	catsper-mi
Caveolinopathies	13	10-31-19	cav
Central Core Disease	3	11-7-19	cco
Charcot-Marie-Tooth Neuropathy Type 1	2	7-5-18	cmt1
Charcot-Marie-Tooth Neuropathy Type 2	2	7-5-18	cmt2
Charcot-Marie-Tooth Neuropathy Type 2E/1F	2	8-15-19	cmt2e
Charcot-Marie-Tooth Neuropathy Type 4	2	7-5-18	cmt4
Charcot-Marie-Tooth Neuropathy Type 4H	2	9-19-19	cmt4h
Charcot-Marie-Tooth Neuropathy Type 4J	2	9-19-19	cmt4j
Charcot-Marie-Tooth Neuropathy X Type 5	15	6-8-23	cmtx5
Congenital Fiber-Type Disproportion	3	4-18-19	cftd
Congenital Hepatic Fibrosis	3	5-14-20	hepatic-fibrosis
Congenital Muscular Dystrophy Overview	4	10-18-18	cmd-overview
Craniofacial Microsomia Overview	7	4-2-20	m-hfm-ov

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Retired *GeneReview* Chapter Title	Reason	Date Retired	SHORTNAME
DFNX1 Nonsyndromic Hearing Loss and Deafness	15	6-8-23	dfnx1
DNM2-Related Intermediate Charcot-Marie-Tooth Neuropathy	2	9-19-19	cmt-dib
Early-Onset Familial Alzheimer Disease	5	9-13-18	alzheimer-early
Episodic Ataxia Type 2	9	10-22-20	ea2
Esophageal Atresia / Tracheoesophageal Fistula Overview	11	4-20-23	tef-ov
Familial Exudative Vitreoretinopathy, Autosomal Dominant	13	9-3-20	fevr
Familial Monosomy 7 Syndrome	11	11-19-20	mono7-mds
Familial Paroxysmal Kinesigenic Dyskinesia	6	1-11-18	pknd
Hereditary Neuralgic Amyotrophy	7	8-29-19	hna
Hirschsprung Disease Overview	12	3-5-20	hirschsprung-ov
Infantile-Onset Spinocerebellar Ataxia	13	5-30-24	sca-io
Leber Congenital Amaurosis	8	5-24-18	lca
Lenz Microphthalmia Syndrome	9	11-7-19	lenz
Leukodystrophy Overview	12	1-30-20	leukodys-ov
Limb-Girdle Muscular Dystrophy Overview	13	5-17-18	lgmd-overview
Majeed Syndrome	14	9-12-19	majeed
Microphthalmia/Anophthalmia/Coloboma Spectrum	4	11-7-19	anophthalmia-ov
Mucolipidosis III Alpha/Beta	10	8-29-19	ml3a
Multiminicore Disease	3	4-18-19	mmd
Myofibrillar Myopathy	3	5-9-19	mfm
Myopathy with Deficiency of ISCU	9, 13	2-10-22	myodef-sda
Myostatin-Related Muscle Hypertrophy	9	4-18-19	mstn
Nemaline Myopathy	3	11-7-19	nem
Neuronal Ceroid-Lipofuscinosis	11	4-11-19	ncl
Nonsyndromic Hearing Loss and Deafness, DFNA3	4	9-7-23	dfna3
Ocular Albinism, X-Linked	13	11-10-21	x-oa
Oculocutaneous Albinism Type 1	4	1-7-21	oca1
Oculocutaneous Albinism Type 2	4	1-7-21	oca2
Optic Atrophy Type 1	4	5-9-24	opa
Polymicrogyria Overview	12	6-6-24	poly
Primary AR Microcephalies and Seckel Syndrome Spectrum Disorders	11	9-10-18	microcephaly
Proopiomelanocortin Deficiency	9	4-16-20	pomc-def

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Retired *GeneReview* Chapter Title	Reason	Date Retired	SHORTNAME
Sialuria	9	3-7-19	sft
Spinocerebellar Ataxia Type 12	9	12-13-18	sca12
Spinocerebellar Ataxia Type 15	4	4-25-24	sca15
Spinocerebellar Ataxia Type 36	9, 13	2-11-21	sca36
Susceptibility to Infection-Induced Acute Encephalopathy 3	9	9-10-20	iiae3
Tetra Amelia	9	3-7-19	tetra-amelia
VCAN-Related Vitreoretinopathy	13	5-16-24	wagner
Wolf-Hirschhorn Syndrome	1	4-18-19	whs
X-Linked Sideroblastic Anemia and Ataxia	9	8-20-20	sider-anemia

1. Non-recurrent deletions or duplications; refers to deletions/duplications of varying size – in contrast to a recurrent deletion/duplication, defined as a deletion/duplication of a specific size (usually mediated by nonallelic homologous recombination) occurring multiple times in the general population
2. Covered in Charcot-Marie-Tooth Hereditary Neuropathy Overview
3. Histologic diagnosis without strong genetic correlation
4. Outdated
5. Covered in Alzheimer Disease Overview
6. Covered in *PRRT2*-Associated Paroxysmal Movement Disorders
7. Rarely genetic
8. Covered in LCA/EOSRD Overview
9. Extremely rare
10. Covered in *GNPTAB*-Related Disorders
11. Chapter does not reflect current use of genetic testing.
12. Phenotype is too broad.
13. Outdated; qualified authors not available for update
14. Covered in *LPIN2*-Related Majeed Syndrome
15. Covered in Phosphoribosylpyrophosphate Synthetase Deficiency
16. Covered in *STRC*-Related Autosomal Recessive Hearing Loss

Note:

- Retired chapters are no longer searchable in *GeneReviews* Advanced Search; however, you can use the following URL and hand-enter the chapter SHORTNAME (see table) to access the archived version of a retired chapter: https://www.ncbi.nlm.nih.gov/books/n/gene/SHORTNAME.
- The information in retired chapters is **for historical reference only** and is potentially misleading as it does not reflect current approaches to diagnosis, management, and genetic counseling. — The Editors

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