

Molecular and phenotypic aspects of *CHD7* muta

American Journal of Medical Genetics, Part A

152A, 674-686

DOI: [10.1002/ajmg.a.33323](https://doi.org/10.1002/ajmg.a.33323)

Citation Report

#	ARTICLE	IF	CITATIONS
1	Inner Ear Anomalies in Congenital Aural Atresia. <i>Otology and Neurotology</i> , 2010, 31, 1421-1426.	0.7	34
2	Chromodomain proteins in development: lessons from CHARGE syndrome. <i>Clinical Genetics</i> , 2010, 78, 11-20.	1.0	63
3	CHD7 functions in the nucleolus as a positive regulator of ribosomal RNA biogenesis. <i>Human Molecular Genetics</i> , 2010, 19, 3491-3501.	1.4	91
4	Mutations in the <i>CHD7</i> Gene: The Experience of a Commercial Laboratory. <i>Genetic Testing and Molecular Biomarkers</i> , 2010, 14, 881-891.	0.3	46
5	Chromatin Remodeling in Development and Disease: Focus on CHD7. <i>PLoS Genetics</i> , 2010, 6, e1001010.	1.5	31
6	Novel CHD7 mutations contributing to the mutation spectrum in patients with CHARGE syndrome. <i>European Journal of Medical Genetics</i> , 2010, 53, 280-285.	0.7	18
7	Clinical genetics of Kallmann syndrome. <i>Annales D'Endocrinologie</i> , 2010, 71, 149-157.	0.6	70
8	CHD7 mutations and CHARGE syndrome: the clinical implications of an expanding phenotype. <i>Journal of Medical Genetics</i> , 2011, 48, 334-342.	1.5	268
9	Genetic Disorders with both Hearing Loss and Cardiovascular Abnormalities. <i>Advances in Oto-Rhino-Laryngology</i> , 2011, 70, 66-74.	1.6	4
10	CHARGE syndrome and CHD7 gene mutation. <i>NeurologÃa (English Edition)</i> , 2011, 26, 255.	0.2	0
11	Mature middle and inner ears express Chd7 and exhibit distinctive pathologies in a mouse model of CHARGE syndrome. <i>Hearing Research</i> , 2011, 282, 184-195.	0.9	36
12	The role of CHD7 and the newly identified WDR11 gene in patients with idiopathic hypogonadotropic hypogonadism and Kallmann syndrome. <i>Molecular and Cellular Endocrinology</i> , 2011, 346, 74-83.	1.6	47
14	CHD7 Mutational Analysis and Clinical Considerations for Auditory Rehabilitation in Deaf Patients with CHARGE Syndrome. <i>PLoS ONE</i> , 2011, 6, e24511.	1.1	25
15	Sox2 cooperates with Chd7 to regulate genes that are mutated in human syndromes. <i>Nature Genetics</i> , 2011, 43, 607-611.	9.4	230
16	Clinical utility gene card for: CHARGE syndrome. <i>European Journal of Human Genetics</i> , 2011, 19, 1017-1017.	1.4	6
17	The DNA-binding domain of the Chd1 chromatin-remodelling enzyme contains SANT and SLIDE domains. <i>EMBO Journal</i> , 2011, 30, 2596-2609.	3.5	104
18	SÃndrome de CHARGE y mutaciÃn en el gen CHD7. <i>NeurologÃa</i> , 2011, 26, 255.	0.3	4
19	Unique phenotype in a patient with CHARGE syndrome. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2011, 2011, 11.	1.6	10

#	ARTICLE	IF	CITATIONS
20	Prevalence of Genetic Testing in CHARGE Syndrome. <i>Journal of Genetic Counseling</i> , 2011, 20, 49-57.	0.9	7
22	VACTERL/VATER Association. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 56.	1.2	336
23	Cranial neural crest cells on the move: Their roles in craniofacial development. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 270-279.	0.7	232
24	Genotype-phenotype analysis of the branchio-oculo-facial syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 22-32.	0.7	61
25	Epigenetic regulation in neural crest development. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2011, 91, 788-796.	1.6	15
26	Reproductive dysfunction and decreased GnRH neurogenesis in a mouse model of CHARGE syndrome. <i>Human Molecular Genetics</i> , 2011, 20, 3138-3150.	1.4	57
27	Antenatal spectrum of CHARGE syndrome in 40 fetuses with <i>CHD7</i> mutations. <i>Journal of Medical Genetics</i> , 2012, 49, 698-707.	1.5	45
28	Chromatin remodeling by the CHD7 protein is impaired by mutations that cause human developmental disorders. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 19238-19243.	3.3	102
29	The Chromatin Fingerprint of Gene Enhancer Elements. <i>Journal of Biological Chemistry</i> , 2012, 287, 30888-30896.	1.6	77
30	Epigenetics and Cardiovascular Development. <i>Annual Review of Physiology</i> , 2012, 74, 41-68.	5.6	187
31	Clinical geneticists' views of VACTERL/VATER association. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 3087-3100.	0.7	78
32	Insight into the Genetic Relevance of Congenital Heart Defects. <i>Current Obstetrics and Gynecology Reports</i> , 2012, 1, 207-215.	0.3	0
33	Congenital Disorders Affecting Sleep. <i>Sleep Medicine Clinics</i> , 2012, 7, 689-702.	1.2	4
34	CHARGE syndrome – A rare combination of cardiac and endocrine disease. <i>International Journal of Cardiology</i> , 2012, 159, 233-234.	0.8	1
35	Duplication of 8q12 encompassing CHD7 is associated with a distinct phenotype but without duane anomaly. <i>European Journal of Medical Genetics</i> , 2012, 55, 646-649.	0.7	9
36	Genetics of Congenital Heart Disease. , 2012, , 473-480.		2
37	Mutation update on the CHD7 gene involved in CHARGE syndrome. <i>Human Mutation</i> , 2012, 33, 1149-1160.	1.1	224
38	A novel classification system to predict the pathogenic effects of CHD7 missense variants in CHARGE syndrome. <i>Human Mutation</i> , 2012, 33, 1251-1260.	1.1	65

#	ARTICLE	IF	CITATIONS
39	Ophthalmic features of CHARGE syndrome with CHD7 mutations. American Journal of Medical Genetics, Part A, 2012, 158A, 514-518.	0.7	24
40	More Clinical Overlap between 22q11.2 Deletion Syndrome and CHARGE Syndrome than Often Anticipated. Molecular Syndromology, 2013, 4, 235-45.	0.3	44
41	Genetics of Female Infertility in Humans. , 2013, , 1-24.		1
43	Knockdown of fbxl10/kdm2bb rescues chd7 morphant phenotype in a zebrafish model of CHARGE syndrome. Developmental Biology, 2013, 382, 57-69.	0.9	34
44	Genetic analysis of the CHD7 gene in Korean patients with CHARGE syndrome. Gene, 2013, 517, 164-168.	1.0	9
45	Brg1 governs distinct pathways to direct multiple aspects of mammalian neural crest cell development. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 1738-1743.	3.3	65
46	Systemic Diagnostic Testing in Patients With Apparently Isolated Uveal Coloboma. American Journal of Ophthalmology, 2013, 156, 1159-1168.e4.	1.7	11
47	Xâ€linked <scp>CHARGE</scp>â€like Abruzzoâ€Erickson syndrome and classic cleft palate withÂankyloglossia result from <i>TBX22</i> splicing mutations. Clinical Genetics, 2013, 83, 352-358.	1.0	22
48	Phenotype in 18 Danish subjects with genetically verified CHARGE syndrome. Clinical Genetics, 2013, 83, 125-134.	1.0	30
49	Structural Pituitary Abnormalities Associated With CHARGE Syndrome. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E737-E743.	1.8	38
50	The role of genetics in the establishment and maintenance of the epigenome. Cellular and Molecular Life Sciences, 2013, 70, 1543-1573.	2.4	53
51	Congenital Heart Defects. , 2013, , 1-51.		2
52	8q12.1q12.3 de novo microdeletion involving the CHD7 gene in a patient without the major features of CHARGE syndrome: Case report and critical review of the literature. Gene, 2013, 513, 209-213.	1.0	8
53	ISWI and CHD Chromatin Remodelers Bind Promoters but Act in Gene Bodies. PLoS Genetics, 2013, 9, e1003317.	1.5	79
54	The Cardiac Phenotype in Patients With a <i>CHD7</i> Mutation. Circulation: Cardiovascular Genetics, 2013, 6, 248-254.	5.1	53
55	Different CHD chromatin remodelers are required for expression of distinct gene sets and specific stages during development of <i>Dictyostelium discoideum</i>. Development (Cambridge), 2013, 140, 4926-4936.	1.2	7
56	Psychomotor and cognitive impairments of children with CHARGE syndrome: Common and variable features. Child Neuropsychology, 2013, 19, 449-465.	0.8	8
57	Outcomes of cochlear implantation in children with CHARGE syndrome. Acta Oto-Laryngologica, 2013, 133, 1148-1153.	0.3	24

#	ARTICLE	IF	CITATIONS
58	Congenital anomalies of kidney and hand: a review. CKJ: Clinical Kidney Journal, 2013, 6, 144-149.	1.4	16
59	Novel Frameshift CHD7 Mutation Related to CHARGE Syndrome. Molecular Syndromology, 2013, 5, 36-40.	0.3	4
61	Kismet Positively Regulates Glutamate Receptor Localization and Synaptic Transmission at the Drosophila Neuromuscular Junction. PLoS ONE, 2014, 9, e113494.	1.1	18
62	Endocrinological Characteristics of 25 Japanese Patients with CHARGE Syndrome. Clinical Pediatric Endocrinology, 2014, 23, 45-51.	0.4	18
63	Family history and clefting as major criteria for CHARGE syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 48-53.	0.7	10
64	Epigenetic regulation in neural crest development. Developmental Biology, 2014, 396, 159-168.	0.9	73
65	The chromatin remodeling protein CHD7, mutated in CHARGE syndrome, is necessary for proper craniofacial and tracheal development. Developmental Dynamics, 2014, 243, 1055-1066.	0.8	34
66	Identification of two novel splice site mutations in <scp>CHD7</scp> gene in two patients with classical and atypical <scp>CHARGE</scp> syndrome phenotype. Clinical Genetics, 2014, 85, 201-202.	1.0	1
67	Symmetrical Chorioretinal Colobomata with Craniovertebral Junction Anomalies in CHARGE Syndrome - A Case Report with Review of Literature. Journal of Clinical Imaging Science, 2014, 4, 5.	0.4	11
68	Genetics of cleft lip and/or cleft palate: Association with other common anomalies. European Journal of Medical Genetics, 2014, 57, 381-393.	0.7	122
69	De novo GLI3 mutation in esophageal atresia: Reproducing the phenotypic spectrum of Gli3 defects in murine models. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1755-1761.	1.8	8
70	Cochlear implantation in children with "CHARGE syndrome": surgical options and outcomes. European Archives of Oto-Rhino-Laryngology, 2014, 271, 489-493.	0.8	20
71	Ocular Involvement in Primary Immunodeficiency Diseases. Journal of Clinical Immunology, 2014, 34, 23-38.	2.0	11
72	Renal Agenesis in Kallmann Syndrome: A Network Approach. Annals of Human Genetics, 2014, 78, 424-433.	0.3	5
73	Epigenetics and Ocular Diseases: From Basic Biology to Clinical Study. Journal of Cellular Physiology, 2014, 229, 825-833.	2.0	23
74	Inappropriate p53 activation during development induces features of CHARGE syndrome. Nature, 2014, 514, 228-232.	13.7	117
75	Genetics of gastrointestinal atresias. European Journal of Medical Genetics, 2014, 57, 424-439.	0.7	17
76	A novel CHD7 mutation in a Chinese patient with CHARGE syndrome. Meta Gene, 2014, 2, 469-478.	0.3	13

#	ARTICLE	IF	CITATIONS
77	CHD7 interacts with BMP R-SMADs to epigenetically regulate cardiogenesis in mice. <i>Human Molecular Genetics</i> , 2014, 23, 2145-2156.	1.4	48
78	Descriptive and risk factor analysis for choanal atresia: The National Birth Defects Prevention Study, 1997â€“2007. <i>European Journal of Medical Genetics</i> , 2014, 57, 220-229.	0.7	25
80	Roles of chromatin remodelers in maintenance mechanisms of multipotency of mouse trunk neural crest cells in the formation of neural crest-derived stem cells. <i>Mechanisms of Development</i> , 2014, 133, 126-145.	1.7	22
81	Cleft Palate in a Mouse Model of <i>SOX2</i> Haploinsufficiency. <i>Cleft Palate-Craniofacial Journal</i> , 2014, 51, 110-114.	0.5	9
82	CHD7 Mutations and CHARGE Syndrome in Semicircular Canal Dysplasia. <i>Otology and Neurotology</i> , 2014, 35, 1466-1470.	0.7	25
83	Cognitiveâ€“motor profile, clinical characteristics and diagnosis of CHARGE syndrome: An Italian experience. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 3042-3051.	0.7	2
84	Terminal 6p deletion syndrome mimicking CHARGE syndrome: A case report. <i>Journal of Pediatric Genetics</i> , 2015, 02, 103-107.	0.3	2
85	CHARGE syndrome due to deletion of region upstream of CHD7 gene START codon. <i>BMC Medical Genetics</i> , 2015, 16, 78.	2.1	6
86	Spontaneous postnatal growth is reduced in children with CHARGE syndrome. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2015, 104, e314-8.	0.7	8
87	Mouse Models for the Dissection of CHD7 Functions in Eye Development and the Molecular Basis for Ocular Defects in CHARGE Syndrome. , 2015, 56, 7923.		26
88	Non-Homologous End Joining Repair Mechanism-Mediated Deletion of <i>CHD7</i> Gene in a Patient with Typical CHARGE Syndrome. <i>Annals of Laboratory Medicine</i> , 2015, 35, 141-145.	1.2	1
89	Epigenetic regulation in the inner ear and its potential roles in development, protection, and regeneration. <i>Frontiers in Cellular Neuroscience</i> , 2014, 8, 446.	1.8	44
90	Genetic Basis of Congenital Heart Disease. <i>NeoReviews</i> , 2015, 16, e340-e350.	0.4	2
91	CHARGE syndrome with oculomotor nerve palsy. <i>Journal of AAPOS</i> , 2015, 19, 555-557.	0.2	5
92	Journey to chew: A case of maxillary duplication and bony syngnathia. <i>Journal of Cranio-Maxillo-Facial Surgery</i> , 2015, 43, 57-61.	0.7	10
93	Epigenetic Developmental Disorders: CHARGE Syndrome, a Case Study. <i>Current Genetic Medicine Reports</i> , 2015, 3, 1-7.	1.9	19
94	CHARGE syndrome: a review of the immunological aspects. <i>European Journal of Human Genetics</i> , 2015, 23, 1451-1459.	1.4	44
95	A critical role for the chromatin remodeller CHD7 in anterior mesoderm during cardiovascular development. <i>Developmental Biology</i> , 2015, 405, 82-95.	0.9	27

#	ARTICLE	IF	CITATIONS
96	Congenital and Acquired Facial Palsies. , 2015, , 225-242.		1
97	Short Children with CHARGE Syndrome: Do They Benefit from Growth Hormone Therapy?. Hormone Research in Paediatrics, 2015, 84, 49-53.	0.8	9
98	Functional Insights into Chromatin Remodelling from Studies on CHARGE Syndrome. Trends in Genetics, 2015, 31, 600-611.	2.9	66
99	Identification of one novel CHD7 mutation in a patient from China with atypical CHARGE syndrome. Gene, 2015, 571, 298-302.	1.0	2
100	Chromo Domain Proteins. , 2016, , 113-125.		0
101	Testicular Disorders. , 2016, , 694-784.		12
102	Cardiovascular Malformations in CHARGE Syndrome with DiGeorge Phenotype: Two Case Reports. Case Reports in Pediatrics, 2016, 2016, 1-6.	0.2	2
103	CT findings of the temporal bone in CHARGE syndrome: aspects of importance in cochlear implant surgery. European Archives of Oto-Rhino-Laryngology, 2016, 273, 4225-4240.	0.8	34
104	The epigenetics of CHARGE syndrome. Frontiers in Biology, 2016, 11, 85-95.	0.7	2
105	CHD associated with syndromic diagnoses: peri-operative risk factors and early outcomes. Cardiology in the Young, 2016, 26, 30-52.	0.4	31
106	Chromatin remodeling enzyme CHD7 is necessary for osteogenesis of human mesenchymal stem cells. Biochemical and Biophysical Research Communications, 2016, 478, 1588-1593.	1.0	12
107	CHD7, Oct3/4, Sox2, and Nanog control FoxD3 expression during mouse neural crest-derived stem cell formation. FEBS Journal, 2016, 283, 3791-3806.	2.2	22
108	Rescue of neural crest-derived phenotypes in a zebrafish CHARGE model by Sox10 downregulation. Human Molecular Genetics, 2016, 25, 3539-3554.	1.4	39
109	A novel frameshift mutation of CHD7 in a Japanese patient with CHARGE syndrome. Human Genome Variation, 2016, 3, 16004.	0.4	8
110	Cochlear Implantation in Patients With CHARGE Syndrome. Annals of Otology, Rhinology and Laryngology, 2016, 125, 924-930.	0.6	7
111	Epigenetics in ENS development and Hirschsprung disease. Developmental Biology, 2016, 417, 209-216.	0.9	36
112	Atypical phenotypes associated with pathogenic CHD7 variants and a proposal for broadening CHARGE syndrome clinical diagnostic criteria. American Journal of Medical Genetics, Part A, 2016, 170, 344-354.	0.7	122
113	Chd7 cooperates with Sox10 and regulates the onset of CNS myelination and remyelination. Nature Neuroscience, 2016, 19, 678-689.	7.1	142

#	ARTICLE	IF	CITATIONS
114	Perspectives in Pediatric Pathology, Chapter 14. Natural History of Undescended Testes. Pediatric and Developmental Pathology, 2016, 19, 183-201.	0.5	5
115	Revealing the function of a novel splice-site mutation of CHD7 in CHARGE syndrome. Gene, 2016, 576, 776-781.	1.0	6
116	Cerebellar vermis hypoplasia in CHARGE syndrome: clinical and molecular characterization of 18 unrelated Korean patients. Journal of Human Genetics, 2016, 61, 235-239.	1.1	13
117	Chromatin deregulation in disease. Chromosoma, 2016, 125, 75-93.	1.0	96
118	Bilateral cochlear nerve absence in a 3 year old child with VACTERL association. International Journal of Pediatric Otorhinolaryngology, 2017, 93, 71-74.	0.4	4
119	Developing a CHARGE syndrome checklist: Health supervision across the lifespan (from head to toe). American Journal of Medical Genetics, Part A, 2017, 173, 684-691.	0.7	32
120	FOXN1 deficient nude severe combined immunodeficiency. Orphanet Journal of Rare Diseases, 2017, 12, 6.	1.2	30
121	Transcribing the connectome: roles for transcription factors and chromatin regulators in activity-dependent synapse development. Journal of Neurophysiology, 2017, 118, 755-770.	0.9	26
122	Accurately annotate compound effects of genetic variants using a context-sensitive framework. Nucleic Acids Research, 2017, 45, e82-e82.	6.5	9
123	Suprameatal Cochlear Implantation in a CHARGE Patient With a Novel CHD7 Variant and KALLMANN Syndrome Phenotype: A Case Report. Otology and Neurotology, 2017, 38, 990-995.	0.7	2
124	Legal Briefs: Should This Neonate with Transposition of the Great Arteries Have Survived?. NeoReviews, 2017, 18, e674-e676.	0.4	0
125	Experiences in feeding and gastrointestinal dysfunction in children with CHARGE syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2947-2953.	0.7	9
126	Maternal residential air pollution and placental imprinted gene expression. Environment International, 2017, 108, 204-211.	4.8	26
127	The Modified Rambo Transcanal Approach for Cochlear Implantation in CHARGE Syndrome. Otology and Neurotology, 2017, 38, 1268-1272.	0.7	7
128	New insights and advances in CHARGE syndrome: Diagnosis, etiologies, treatments, and research discoveries. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 397-406.	0.7	46
129	Phenotype and genotype analysis of a French cohort of 119 patients with CHARGE syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 417-430.	0.7	65
130	Reproductive endocrine phenotypes relating to <i>CHD7</i> mutations in humans. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 507-515.	0.7	41
131	CHARGE Syndrome. , 2017, , 407-415.		0

#	ARTICLE	IF	CITATIONS
132	Modeling Syndromic Congenital Heart Defects in Zebrafish. <i>Current Topics in Developmental Biology</i> , 2017, 124, 1-40.	1.0	36
133	Individual Clinically Diagnosed with CHARGE Syndrome but with a Mutation in KMT2D, a Gene Associated with Kabuki Syndrome: A Case Report. <i>Frontiers in Genetics</i> , 2017, 8, 210.	1.1	18
134	Chd7 Is Critical for Early T-Cell Development and Thymus Organogenesis in Zebrafish. <i>American Journal of Pathology</i> , 2018, 188, 1043-1058.	1.9	28
135	Cleft Lip and Palate in CHARGE Syndrome. <i>Cleft Palate-Craniofacial Journal</i> , 2018, 55, 342-347.	0.5	9
136	Chromatin remodeler CHD7 regulates the stem cell identity of human neural progenitors. <i>Genes and Development</i> , 2018, 32, 165-180.	2.7	28
137	Epigenetic crosstalk: Pharmacological inhibition of HDACs can rescue defective synaptic morphology and neurotransmission phenotypes associated with loss of the chromatin reader Kismet. <i>Molecular and Cellular Neurosciences</i> , 2018, 87, 77-85.	1.0	14
138	Dysregulation of cotranscriptional alternative splicing underlies CHARGE syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E620-E629.	3.3	28
139	Differentiation potential of Pluripotent Stem Cells correlates to the level of CHD7. <i>Scientific Reports</i> , 2018, 8, 241.	1.6	20
140	Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. <i>American Journal of Human Genetics</i> , 2018, 102, 156-174.	2.6	135
141	Enhancers: bridging the gap between gene control and human disease. <i>Human Molecular Genetics</i> , 2018, 27, R219-R227.	1.4	51
142	Transcriptional and Epigenetic Control of Mammalian Olfactory Epithelium Development. <i>Molecular Neurobiology</i> , 2018, 55, 8306-8327.	1.9	25
143	Clinical data and genetic mutation in Kallmann syndrome with CHARGE syndrome. <i>Medicine (United Tj ETQq1 1 0.784314 rgBT /Ove</i>	0.4	7
144	Kat2a and Kat2b Acetyltransferase Activity Regulates Craniofacial Cartilage and Bone Differentiation in Zebrafish and Mice. <i>Journal of Developmental Biology</i> , 2018, 6, 27.	0.9	32
145	Transcriptome analysis of <i>Xenopus</i> orofacial tissues deficient in retinoic acid receptor function. <i>BMC Genomics</i> , 2018, 19, 795.	1.2	8
146	Genetic Basis for Congenital Heart Disease: Revisited: A Scientific Statement From the American Heart Association. <i>Circulation</i> , 2018, 138, e653-e711.	1.6	387
147	Identifying the genetic causes for prenatally diagnosed structural congenital anomalies (SCAs) by whole-exome sequencing (WES). <i>BMC Medical Genomics</i> , 2018, 11, 93.	0.7	32
148	ALS and CHARGE syndrome: a clinical and genetic study. <i>Acta Neurologica Belgica</i> , 2018, 118, 629-635.	0.5	4
149	Syndromic hearing loss molecular diagnosis: Application of massive parallel sequencing. <i>Hearing Research</i> , 2018, 370, 181-188.	0.9	6

#	ARTICLE	IF	CITATIONS
150	Neurocristopathies: New insights 150 years after the neural crest discovery. <i>Developmental Biology</i> , 2018, 444, S110-S143.	0.9	136
151	Functional Analyses of a Novel Splice Variant in the CHD7 Gene, Found by Next Generation Sequencing, Confirm Its Pathogenicity in a Spanish Patient and Diagnose Him with CHARGE Syndrome. <i>Frontiers in Genetics</i> , 2018, 9, 7.	1.1	19
152	Atypical CHARGE associated with a novel frameshift mutation of CHD7 in a Chinese neonatal patient. <i>BMC Pediatrics</i> , 2018, 18, 203.	0.7	4
153	Systematic review of cochlear implantation in CHARGE syndrome. <i>Cochlear Implants International</i> , 2019, 20, 266-280.	0.5	12
154	Auditory Skills following Cochlear Implantation in Children with the Charge Syndrome. <i>Audiology and Neuro-Otology</i> , 2019, 24, 139-146.	0.6	3
155	Fetal Cardiovascular Disease. , 2019, , 252-282.		0
156	A novel CHD7 variant disrupting acceptor splice site in a patient with mild features of CHARGE syndrome: a case report. <i>BMC Medical Genetics</i> , 2019, 20, 127.	2.1	7
157	Sema3E is required for migration of cranial neural crest cells in zebrafish: Implications for the pathogenesis of CHARGE syndrome. <i>International Journal of Experimental Pathology</i> , 2019, 100, 234-243.	0.6	15
158	Relative Preservation of Superior Semicircular Canal Architecture in CHARGE Syndrome. <i>Otolaryngology - Head and Neck Surgery</i> , 2019, 160, 1095-1100.	1.1	2
159	Fetal MRI findings in a retrospective cohort of 26 cases of prenatally diagnosed CHARGE syndrome individuals. <i>Prenatal Diagnosis</i> , 2019, 39, 781-791.	1.1	15
160	De Novo Duplication in the CHD7 Gene Associated With Severe CHARGE Syndrome. <i>Genomics Insights</i> , 2019, 12, 117863101983901.	3.0	1
161	Developmental processes regulate craniofacial variation in disease and evolution. <i>Genesis</i> , 2019, 57, e23249.	0.8	13
162	Optic Disc Coloboma in children – prevalence, clinical characteristics and associated morbidity. <i>Acta Ophthalmologica</i> , 2019, 97, 478-485.	0.6	7
164	The role of p53 in developmental syndromes. <i>Journal of Molecular Cell Biology</i> , 2019, 11, 200-211.	1.5	51
165	Relationship between neural crest cell specification and rare ocular diseases. <i>Journal of Neuroscience Research</i> , 2019, 97, 7-15.	1.3	42
166	Chemical screens in a zebrafish model of CHARGE syndrome identifies small molecules that ameliorate disease-like phenotypes in embryo. <i>European Journal of Medical Genetics</i> , 2020, 63, 103661.	0.7	10
167	Aggressive-like behavior and increased glycine transporters in a zebrafish model of CHARGE syndrome. <i>Behavioural Brain Research</i> , 2020, 378, 112293.	1.2	6
168	Nonneoplastic Diseases of the Testis. , 2020, , 549-730.e81.		2

#	ARTICLE	IF	CITATIONS
169	A new imaging entity consistent with partial ectopic posterior pituitary gland: report of six cases. <i>Pediatric Radiology</i> , 2020, 50, 107-115.	1.1	5
170	Phenotypic Spectrum of Idiopathic Hypogonadotropic Hypogonadism Patients With <i>CHD7</i> Variants From a Large Chinese Cohort. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 1515-1526.	1.8	12
171	Identification of a novel heterozygous missense mutation of <i>SEMA3E</i> (c.1327C>A; p. Ala443Thr) in a labor induced fetus with CHARGE syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1034.	0.6	4
172	Congenital Heart Defects. , 2020, , 3-75.		2
173	Congenital heart defects in CHARGE: The molecular role of <i>CHD7</i> and effects on cardiac phenotype and clinical outcomes. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 81-89.	0.7	17
174	The mechanisms of action of chromatin remodelers and implications in development and disease. <i>Biochemical Pharmacology</i> , 2020, 180, 114200.	2.0	18
175	Genetics of syndromic ocular coloboma: CHARGE and COACH syndromes. <i>Experimental Eye Research</i> , 2020, 193, 107940.	1.2	23
176	<i>CHD7</i> regulates cardiovascular development through ATP-dependent and -independent activities. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 28847-28858.	3.3	27
177	<i>CHARGE</i> syndrome without colobomas: Ophthalmic findings. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 611-617.	0.7	4
178	Molecular Insights Into the Causes of Human Thymic Hypoplasia With Animal Models. <i>Frontiers in Immunology</i> , 2020, 11, 830.	2.2	16
179	<i>CHD7</i> missense variants and clinical characteristics of Chinese males with infertility. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1372.	0.6	3
180	Defects in thymic development. , 2020, , 357-379.		2
181	A case series of CHARGE syndrome: identification of key features for a neonatal diagnosis. <i>Italian Journal of Pediatrics</i> , 2020, 46, 53.	1.0	6
182	CHARGE syndrome in the era of molecular diagnosis: Similar outcomes in those without coloboma or choanal atresia. <i>European Journal of Medical Genetics</i> , 2021, 64, 104103.	0.7	4
184	Using an aquatic model, <i>Xenopus laevis</i> , to uncover the role of chromodomain 1 in craniofacial disorders. <i>Genesis</i> , 2021, 59, e23394.	0.8	10
186	CHARGE Syndrome. , 2021, , 1-7.		0
187	The Malformed Eye. , 2021, , 239-274.		0
188	Whole Exome Sequencing in Coloboma/Microphthalmia: Identification of Novel and Recurrent Variants in Seven Genes. <i>Genes</i> , 2021, 12, 65.	1.0	16

#	ARTICLE	IF	CITATIONS
189	Otopathologic Abnormalities in CHARGE Syndrome. <i>Otolaryngology - Head and Neck Surgery</i> , 2022, 166, 363-372.	1.1	6
190	A framework for the evaluation of patients with congenital facial weakness. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 158.	1.2	3
191	Semaphorin Regulation by the Chromatin Remodeler CHD7: An Emerging Genetic Interaction Shaping Neural Cells and Neural Crest in Development and Cancer. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 638674.	1.8	5
192	Antenatal diagnosis of CHARGE syndrome: Prenatal ultrasound findings and crucial role of fetal dysmorphic signs. About a series of 10 cases and review of literature. <i>European Journal of Medical Genetics</i> , 2021, 64, 104189.	0.7	8
193	Chromatin remodeller CHD7 is required for GABAergic neuron development by promoting PAQR3 expression. <i>EMBO Reports</i> , 2021, 22, e50958.	2.0	15
194	Epigenetic Regulation of Cardiac Neural Crest Cells. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 678954.	1.8	6
195	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. <i>Human Genetics</i> , 2021, 140, 1109-1120.	1.8	18
196	The CHD8/CHD7/Kismet family links blood-brain barrier glia and serotonin to ASD-associated sleep defects. <i>Science Advances</i> , 2021, 7, .	4.7	24
197	Bilateral Choanal Atresia and Endoscopic Surgery: A Chance for CHARGE Patients. <i>Journal of Clinical Medicine</i> , 2021, 10, 2951.	1.0	1
198	The Role of De Novo Variants in Formation of Human Anorectal Malformations. <i>Genes</i> , 2021, 12, 1298.	1.0	1
199	Persistent Trigeminal Artery: A Novel Imaging Finding in CHARGE Syndrome. <i>American Journal of Neuroradiology</i> , 2021, 42, 1898-1903.	1.2	0
200	A Novel N-terminal Region to Chromodomain in CHD7 is Required for the Efficient Remodeling Activity. <i>Journal of Molecular Biology</i> , 2021, 433, 167114.	2.0	4
202	Primary and Secondary Hypogonadism. <i>Endocrinology</i> , 2017, , 687-747.	0.1	6
203	Pediatric Cranial Neuropathies. , 2017, , 181-198.		1
204	Testicular Disorders. , 2011, , 688-777.		15
205	Otitis Media in a New Mouse Model for CHARGE Syndrome with a Deletion in the Chd7 Gene. <i>PLoS ONE</i> , 2012, 7, e34944.	1.1	23
206	Growth hormone deficiency as a cause for short stature in Wiedemann-Steiner Syndrome. <i>Endocrinology, Diabetes and Metabolism Case Reports</i> , 2018, 2018, .	0.2	14
207	CHARGE syndrome associated with de novo (I1460Rfs*15) frameshift mutation of CHD7 gene in a patient with arteria lusoria and horseshoe kidney. <i>Experimental and Therapeutic Medicine</i> , 2020, 20, 479-485.	0.8	12

#	ARTICLE	IF	CITATIONS
208	A case of CHARGE syndrome featuring immunodeficiency and hypocalcemia. <i>Journal of Genetic Medicine</i> , 2015, 12, 57-60.	0.1	1
209	CHARGE syndrome modeling using patient-iPSCs reveals defective migration of neural crest cells harboring CHD7 mutations. <i>ELife</i> , 2017, 6, .	2.8	52
210	HERVH-derived lncRNAs negatively regulate chromatin targeting and remodeling mediated by CHD7. <i>Life Science Alliance</i> , 2022, 5, e202101127.	1.3	3
211	A Balancing Act: p53 Activity from Tumor Suppression to Pathology and Therapeutic Implications. <i>Annual Review of Pathology: Mechanisms of Disease</i> , 2022, 17, 205-226.	9.6	13
212	CHARGE Syndrome. , 2012, , 323-330.		0
213	Molecular confirmation of CHARGE syndrome from umbilical cord blood stem cells from a death newborn and identification of a new mutation in the exon 29 of the <i>CHD7</i> gene. <i>Stem Cell Discovery</i> , 2012, 02, 1-4.	0.5	0
215	The Malformed Eye. , 2014, , 231-259.		0
216	Genetics of Congenital and Acquired Cardiovascular Disease. , 2014, , 3-16.		1
217	Delayed Puberty. , 2014, , 203-217.		1
218	CHARGE Syndrome. , 2016, , 1-9.		0
219	Charge Syndrome. <i>Pondicherry Journal of Nursing</i> , 2018, 11, 30-33.	0.0	0
220	Primary and Secondary Hypogonadism. <i>Endocrinology</i> , 2017, , 1-62.	0.1	0
221	CHARGE Syndrome, Esophagus. <i>Encyclopedia of Pathology</i> , 2017, , 123-127.	0.0	0
222	Charge syndrome: Clinical characteristics and elements of treatment. <i>Timocki Medicinski Glasnik</i> , 2019, 44, 124-129.	0.0	0
223	Targeted next-generation sequencing for research and diagnostics in congenital heart disease, and cleft lip and/or palate. <i>Molecular Medicine Reports</i> , 2019, 19, 3831-3840.	1.1	7
224	CHARGE Syndrome. , 2020, , 1-7.		1
225	Syndromic Eye Anomalies. , 2020, , 41-95.		0
226	The chromatin remodelling factor Chd7 protects auditory neurons and sensory hair cells from stress-induced degeneration. <i>Communications Biology</i> , 2021, 4, 1260.	2.0	10

#	ARTICLE	IF	CITATIONS
228	Phenotypic spectrum of patients with mutations in CHD7: clinical implications of endocrinological findings. <i>Endocrine Connections</i> , 2022, 11, .	0.8	4
229	Analyzing the effect of deleterious non-synonymous SNPs causing CHARGE syndrome associated with the CHD7 protein using computational approaches. <i>Journal of Proteins and Proteomics</i> , 2022, 13, 63.	1.0	2
230	De novo Splice Site Mutation of the <i>CHD7</i> Gene in a Chinese Patient with Typical CHARGE Syndrome. <i>Orl</i> , 2022, 84, 417-424.	0.6	3
232	Facial Asymmetry: A Narrative Review of the Most Common Neurological Causes. <i>Symmetry</i> , 2022, 14, 737.	1.1	3
235	CHARGE syndrome in children with congenital choanal atresia. <i>Vestnik Otorinolaringologii</i> , 2022, 87, 7.	0.0	0
236	Growth hormone deficiency in a boy with Wiedemann-Steiner syndrome: a case report and review. <i>Annals of Pediatric Endocrinology and Metabolism</i> , 2023, 28, S25-S28.	0.8	2
237	Whole-exome sequencing identified five novel <i>de novo</i> variants in patients with unexplained intellectual disability. <i>Journal of Clinical Laboratory Analysis</i> , 0, , .	0.9	1
238	Treatment of choanal atresia in a cohort of 29 patients: Determinants for success or failure. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2022, 160, 111240.	0.4	1
240	Genética de las fisuras labiopalatinas: una revisión general de factores ambientales. <i>Revista Med</i> , 2022, 29, 93-106.	0.1	1
242	Eyes on CHARGE syndrome: Roles of CHD7 in ocular development. <i>Frontiers in Cell and Developmental Biology</i> , 0, 10, .	1.8	4
243	A clinical case of surgical treatment of complicated cataract in a patient with Hall-Hittner syndrome. <i>Journal of Clinical Practice</i> , 0, , .	0.2	0
244	CHD7 regulates otic lineage specification and hair cell differentiation in human inner ear organoids. <i>Nature Communications</i> , 2022, 13, .	5.8	19
245	Chromatin remodeler Chd7 regulates photoreceptor development and outer segment length. <i>Experimental Eye Research</i> , 2023, 226, 109299.	1.2	0
246	The Incidence and Outcomes for Children with Cleft Palate and/or Lip and CHARGE Syndrome. <i>Cleft Palate-Craniofacial Journal</i> , 0, , 105566562211345.	0.5	0
247	Acute myeloid leukemia associated with <i>CHARGE</i> syndrome. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	0
248	Patterns of co-occurring birth defects in children with anotia and microtia. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	1
249	Morphological and sensorimotor phenotypes in a zebrafish <i>CHARGE</i> syndrome model are domain-dependent. <i>Genes, Brain and Behavior</i> , 2023, 22, .	1.1	3
250	SIN3A Defects Associated with Syndromic Congenital Hypogonadotropic Hypogonadism: An Overlap with Witteveen-Kolk Syndrome. <i>Neuroendocrinology</i> , 2023, 113, 834-843.	1.2	2

#	ARTICLE	IF	CITATIONS
251	ATM-dependent phosphorylation of CHD7 regulates morphogenesis-coupled DSB stress response in fetal radiation exposure. <i>Molecular Biology of the Cell</i> , 2023, 34, .	0.9	0
253	The NuRD Complex in Neurodevelopment and Disease: A Case of Sliding Doors. <i>Cells</i> , 2023, 12, 1179.	1.8	2
257	Optic Nerve Coloboma: A Boy with Poor Vision and Microphthalmos. , 2023, , 7-10.		0
262	Advance and Application of Single-cell Transcriptomics in Auditory Research. <i>Neuroscience Bulletin</i> , 0, , .	1.5	1