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ARTICLE

DOORS Syndrome: Phenotype, Genotype and Comparison With Coffin-Siris Syndrome

PHILIPPE M. CAMPEAU, RAOUL C. HENNEKAM, AND THE DOORS SYNDROME COLLABORATIVE GROUP

DOORS syndrome (Deafness, Onychodystrophy, Osteodystrophy, mental Retardation, Seizures) is characterized mainly by sensorineural deafness, shortened terminal phalanges with small nails of hands and feet, intellectual deficiency, and seizures. Half of the patients with all clinical features have mutations in *TBC1D24*. We review here the manifestations of patients clinically diagnosed with DOORS syndrome. In this cohort of 32 families (36 patients) we detected 13 individuals from 10 families with *TBC1D24* mutations. Subsequent whole exome sequencing in the cohort showed the same de novo*SMARCB1* mutation (c.1130G>A), known to cause Coffin-Siris syndrome, in two patients. Distinguishing features include retinal anomalies, Dandy-Walker malformation, scoliosis, rocker bottom feet, respiratory difficulties and absence of seizures, and 2-oxoglutaric aciduria in the patients with the *SMARCB1* mutation. We briefly discuss the heterogeneity of the DOORS syndrome phenotype and the differential diagnosis of this condition. © 2014 Wiley Periodicals, Inc.

KEY WORDS: DOOR syndrome; DOORS syndrome; phenotype; deafness; seizures; intellectual disability; TBC1D24; SMARCB1; genotype-phenotype correlation

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INTRODUCTION

The acronym DOOR syndrome was first used by the American pediatrician Ronald Cantwell describing a 14-year-old girl born to first cousin parents, with a short distal phalanges of fingers and toes, small nails, tri-phalangeal thumbs, deafness and intellectual disability [Cantwell, 1975]. He remarked very similar cases were reported before [Feinmesser and Zelig, 1961; Goodman et al., 1969; Walbaum et al., 1970], yet the digital findings varied and also the pattern of inheritance seemed to vary. Qazi and Nangia suggested the use of the acronym DOORS syndrome because most individuals also have seizures [Qazi and Nangia, 1984]. DOORS syndrome has also been referred to as digito-reno-cerebral syndrome [Eronen et al., 1985; Winter, 1993], or Eronen syndrome [Le Merrer et al., 1992]. Thirty-two cases were reviewed by James and co-workers in 2007 [James et al., 2007]. Additional signs and symptoms were recognized such as optic neuropathy, abnormalities on brain Magnetic Resonance Imaging, peripheral neuropathy, and a large and broad nasal ridge and tip, and broad nasal base as the most common facial characteristics.

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In a joint project between Houston, London, and Amsterdam we enrolled patients from 26 families in a whole exome sequencing project, and identified *TBC1D24* mutations in half of the patients with all five main features of the

Conflict of interest: none

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disease [Campeau et al., 2014]. We have subsequently enrolled another six patients from six families and through additional whole exome sequencing we identified two individuals with the same de novo *SMARCB1* mutation which has earlier been reported to cause Coffin-Siris syndrome [Tsurusaki et al., 2012].

In the present review we describe the phenotype of 13 individuals with a *TBC1D24* mutation, the two patients with a *SMARCB1* mutation, the 23 patients in whom no mutation could be detected in these genes, and compare the phenotypes of these groups of patients. Families with dominant deafness-onychodystrophy (DDOD) syndrome will be reported elsewhere [Campeau et al., 2014].

METHODS

All individuals who participate in the present study have three or more of the five main features of DOORS syndrome (deafness; abnormal nails of hands and/or feet; abnormal digits in the hands and/or feet; developmental delay or intellectual disability; seizures). We contacted physicians who had published cases and previous collaborators for further detailed information. Available clinical pictures and radiographs were evaluated. The molecular data were retrieved from our earlier publication [Campeau et al., 2014]. Whole exome sequencing was performed as described in the former publication.

RESULTS

Genotypes

Mutations in *TBC1D24* had been identified initially in various epileptic syndromes, such as focal epilepsy with intellectual disability [Corbett et al., 2010], familial infantile myoclonic epilepsy [Falace et al., 2010], myoclonic epilepsy with dystonia [Guven and Tolun, 2013], and familial malignant migrating partial seizures of infancy [Milh et al., 2013]. By analyzing the results of whole exome sequencing in 17 of (at that time) 26 families (30 patients) with DOORS syndrome, we found eight different *TBC1D24* mutations in nine. All 9 belonged to the 18 families in which the affected children have all five cardinal features of DOORS syndrome. In these families, DOORS syndrome was transmitted in an autosomal recessive fashion. Subsequent to our report, different mutations were identified in autosomal recessive non-syndromic deafness (DFNB86, mutations c.208G > T, p.(Asp70Tyr) and c.878G > C, p.(Arg293Pro)) [Rehman et al., 2014], and autosomal dominant non-syndromic deafness (mutation c.533C > T, p.(Ser178Leu)) [Azaiez et al., 2014; Zhang et al., 2014]. The various conditions named above are each caused by separate mutations, suggesting until now there is a clear genotype-phenotype correlation in *TBC1D24*-related conditions (see Figure 1).

Since our previous report, we have enrolled six new families in our study, and have continued exome sequencing and analysis in our cohort. We identified two more *TBC1D24* mutations in one affected child (c.1460dupA, p.(His487Glnfs*71) and c.313T > C p.(Cys105Arg)). When analyzing rare variants in common genes between affected individuals, we identified two patients with a de novo *SMARCB1* c.1130G > A, p.Arg377His mutation, confirmed by Sanger sequencing.

Phenotype in Patients With TBC1D24 Mutations

The phenotype of the present cohort of individuals with clinically diagnosed DOORS syndrome and a *TBC1D24* mutation is to a larger extent homogeneous (Table I). Surely this is in part explainable by the bias introduced at the selection for molecular analysis. It might be that in evaluating larger series of individuals with intellectual disability for variants in large series of genes that all can cause intellectual disability (targeted whole exome sequencing) variants will also be detected in individuals who have a much less clear phenotype or even a completely different phenotype.

The phenotype of the present DOORS syndrome series with *TBC1D24* mutations shows that in 100% of the cases they have intellectual disability or developmental delay, seizures, deafness, short distal phalanges, and small or absent nails (Table I). The majority presents with an increased urinary 2-oxoglutaric acid excretion as well. Prenatal or postnatal growth is usually not disturbed although it can occur, and also skull growth is impaired only in a quarter of the patients. The facial characteristics can be quite different; only a broad nasal bridge is present in half of the cases but other signs are present in single individuals only (Fig. 2 a), except for a high palate and thick gingiva which each were present in two cases. One patient had a craniosynostosis of the sagittal suture. Next to the small distal phalanges the acral manifestations consist mainly of triphalangeal thumbs, present in a quarter of the present series. Internal organs are usually normal although one case had a complex heart malformation and two had renal anomalies. MRIs of the brain show abnormalities in half of the present case series, and findings vary from a thin cortex, and subdural effusions to increased T2 signals and a delayed myelination.

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Phenotype in Patients With SMARCB1 Mutations

Analysis of the clinical features of the two patients with a clinical diagnosis of DOORS syndrome in whom a

TABLE I. A Summary of All Main Clinical Findings in 38 Individuals Clinically Diagnosed With DOORS Syndrome, Subgrouped by Results of Molecular Studies, and Compared to Findings in *SMARCB1*-positive Coffin-Siris Patients

		Patients suspected of having DOORS syndrome			Coffin-Siris syndrome, *SMARCB1* positive (n = 9)[b]
		TBC1D24 positive (n = 13)	*SMARCB1* positive (n = 2)[a]	*TBC1D24/SMARCB1* negative (n = 23)	
Growth	IUGR/SGA	0/12	2/2	5/23	3/5
	Failure to Thrive	1/12	1/1	8/23	2/4
Neurology	Microcephaly	3/12	1/2	5/23	5/8
	Delay cognition/development[c]	13/13 (3+++/1+/9 nos)	1/1 (1+++/1 nos)	23/23 (10+++/13 nos)	9/9 (4+++/5 nos)
	Hypotonia	N./A	2/2	N/A	7/9
	Seizures	13/13	0/2	12/23	6/9
	MRI findings	2 TC; 3 iT2; 1 SE; 1 DM	2/2 DW	2 AC; 2 DM; 1 FA; 2 SC; 1 ACy; 1VS	5/6 AC
Senses	Hearing loss	13/13	2/2	12/23	5/8
	Eye anomalies	2 My; 1 AR	1RC; 1 MG	1 My; 2 S; 1 RC; 3 MVT	5/8 My
Cranio-facial	Coarse face	0/12	1/2	3/23	5/5
	Sparse scalp hair	0/12	2/2	3/23	8/9
	Narrow bifrontal diameter	1/12	2/2	2/23	N/A
	Bushy eyebrows	0/12	1/2	1/23	9/9
	Broad nasal bridge	6/12	2/2	7/23	4/5
	Thick vermilions	0/12	1/2	0/23	7/9
	Wide mouth	0/12	2/2	1/23	6/9
	High-arched palate	2/12	2/2	8/23[d]	5/5
	Thick gingiva	2/12	0/2	2/23	N/A
Musculoskeletal	Short neck	0/12	2/2	1/23	N/A
	Spinal anomalies	0/12	2/2 S	2/23	4/5
	Small distal phalanges	13/13	2/2	19/23	1/4
	Small 5th finger	12/12	2/2	19/23	4/5
	Triphalangeal thumb	3/12	0/2	9/23	N/A
	Long/broad halluces	0/12	2/2	0/23	N/A[e]
	Feet	1/12 DC	2/2 RBF	0/23	N/A
Ectoderm	Small/absent nails	13/13	2/2	23/23	8/9
Other	Renal anomalies	1 Hy; 1 NeCa	1/2 Hy	3/23	2/8 HK
	Cardiac malformations	1 DORV	1 PDA+PFO; 1 ASD+TR+ LSVC	1PDA+ VSD; 2ASD; 1 DORV; 1 DCRV; 1CoA	4/9 De
	Abn 2-oxoglutarate excretion	7/11	0/2[f]	12/23	N/A

TC, thin cortex; iT2, increased T2 signal; SE, subdural effusion; DM, delayed myelination; AC, absent/small callosal body; DW, Dandy-Walker malformation; FA, falx agenesis; SC, small cerebellum/vermis; ACy, arachnoid cyst; VS, enlarged Virchow-Robin spaces; My, myopia; AR, absent light response; S, strabism; HM, hypermetropia; RC, retina coloboma; MG, morning glory; MVT, abnormal movements such as nystagmus; S, scoliosis; DC, deformed calcaneus; RBC, rocker bottom feet; Hy, hydronephrosis; NeCa, nephrocalcinosis; HK, horseshoe kidney; DORV, double outlet right ventricle; PDA, persistent ductus arteriosus; PFO, persistent foramen ovale; ASD, atrial septal defect; TR, tricuspid regurgitation; LSVC, left superior vena cava draining to coronary sinus; VSD, ventricular septal defect; CoA, coarctation aortae; DCRV, Double chambered right ventricle; De, dextrocardia.

[a]One of these patients died in the neonatal period.

[b]From Santen et al. [2013] and Tsurusaki et al. [2012].

[c]+ mild (IQ50-69); ++ moderate (IQ35-49); +++ severe (IQ<35); ? cognitive delay but severity not specified.

[d]Three of them had cleft palate.

[e]Prominent distal phalanges in 3.

[f]in one increased lactate excretion.

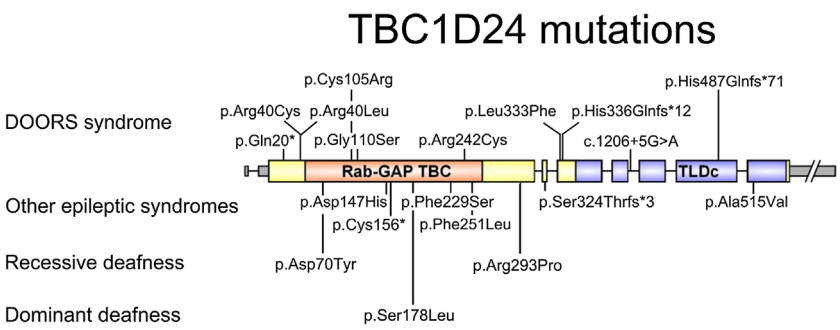

Figure 1. Graph of *TBC1D24* and the mutations described in DOORS syndrome, other epileptic syndromes and autosomal recessive and autosomal dominant deafness.

SMARCAB1 mutation was found is hampered by the small numbers, and the fact that one of the two patients died in the first year of life (Table I). In comparison with the DOORS syndrome cohort caused by *TBC1D24* mutations all major manifestations were present in both patients, except for the epilepsy. We cannot exclude however that epilepsy would still develop in one patient or would have developed in the other if this patient had not died at an early age. There are differences however: both patients with the *SMARCB1* mutation have retinal anomalies (colobomas in one, morning glory anomaly in the other), both have a Dandy-Walker malformation, and also a short neck, scoliosis, and respiratory difficulties occur in both. In the limbs the long and broad halluces and rocker bottom feet are present in both. They do not have an 2-oxoglutaric aciduria. The facial signs seem to differ as well: sparse hair, narrow bi-frontal diameters, a broad nasal bridge, and wide mouth, all together giving rise to a somewhat 'coarse' face were present (Fig. 2b).

If the two patients are compared to nine patients with *SMARCB1* mutations reported recently [Santen et al., 2013; Tsurusaki et al., 2014], there is an obvious overlap, but they are not exactly the same (Table I), especially with respect to brain and eye signs, shape of the halluces, and presence of cardiac malformations. The number of patients is very small though, and this cannot be concluded with any certainty. It remains difficult how to classify these patients [Hennekam, 2007]: should these patients be reclassified as patients with Coffin-Siris syndrome or is the clinical diagnosis of DOORS syndrome still justified? We suggest at the present to leave this undecided, until larger groups of individuals with a DOORS phenotype have been investigated on a molecular level, and it will become clear whether there is a group of individuals with genuine DOORS characteristics who have a *SMARCB1* mutation, or whether within the group of Coffin-Siris patients some show a less clear Coffin-Siris phenotype but still have sufficient signs and symptoms to be classified as such, and have a *SMARCB1* mutation.

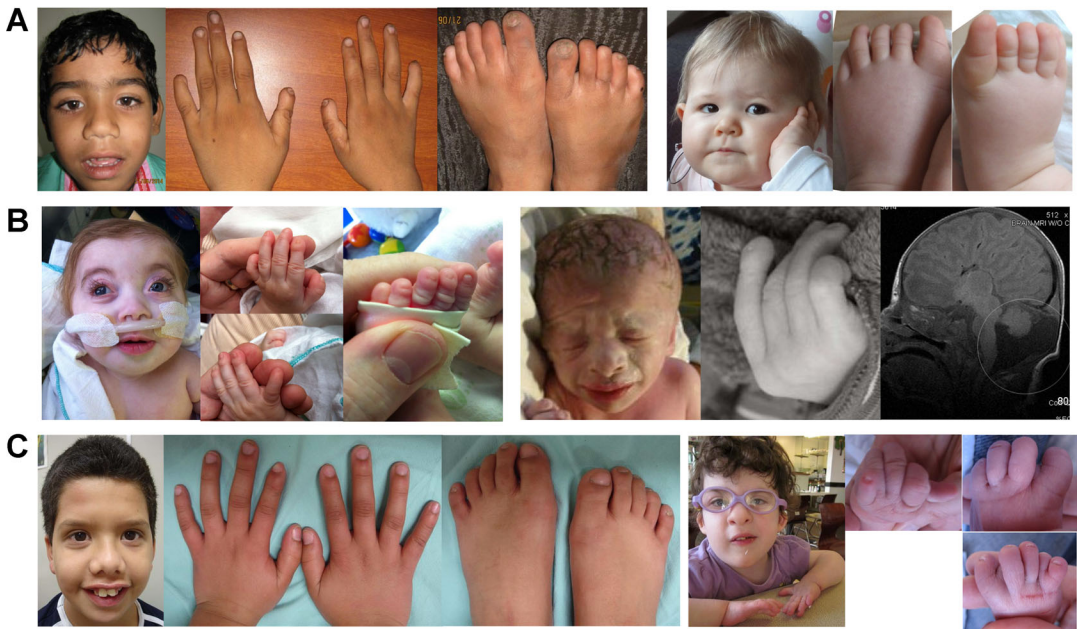

Figure 2. Clinical pictures of faces and limbs of a selection of the presently described individuals with clinically diagnosed DOORS syndrome. **A**: Individuals with a *TBC1D24* mutation; (**B**). The two individuals with a *SMARCB1* mutation, including an MRI showing the Dandy-Walker malformation; (**C**), Two individuals without detectable *TBC1D24* and *SMARCB1* mutations.

Phenotype of Patients Without TBC1D24 or SMARCB1 Mutations

We have evaluated the phenotype of a cohort of 23 individuals with a clinical diagnosis DOORS syndrome in whom neither *TBC1D24* nor *SMARCAB1* mutation analysis yielded an abnormality. As can be expected the phenotype of these patients is variable (Table I; Fig. 2c), as it seems likely that this group will not be genetically homogeneous. The core DOORS characteristics are obviously frequent in this group due to the inclusion criteria. Still, the frequency of hearing loss (12/23) and seizures (12/23) are not as high as in the group with a *TBC1D24* mutation. The other main characteristics of DOORS syndrome occur in all, and also ophthalmological and MRI findings, and excretion of 2-oxoglutarate are not essentially different in nature and frequency. Further differences are the more common prenatal (5/23) and postnatal (8/23) growth disturbance, and possibly the occurrence of tri-phalangeal thumbs is higher as well. The facial signs are variable, like in the *TBC1D24* positive cases.

We assume that further careful phenotyping on one hand, and comparisons of data from whole exome sequencing of affected individuals (and if available their parents) on the other hand, will allow recognition of other gene(s) causing DOORS syndrome, and such studies are at present in progress.

CONCLUSION

DOORS syndrome should be differentiated from entities caused by disturbances in the BAF complex such as Coffin-Siris syndrome [Tsurusaki et al., 2014] and Nicolaides-Baraitser syndrome [Sousa et al., 2009; Van Houdt et al., 2012], but also from other entities without a known cause as Temple-Baraitser syndrome [Jacquinet et al., 2010], and Zimmerman-Laband syndrome [Castori et al., 2013]. The core characteristic of DOORS syndrome can occur in each of these, and often it will depend on the presence of additional characteristics whether these related entities can be recognized. It may be considered to add to the 5 core name giving characteristics of DOORS also absence of other major malformations or significant dysmorphic characteristics as criteria for the diagnosis. However, the number of individuals with DOORS syndrome in whom the diagnosis can be confirmed molecularly is still small, and with this also the variability remains uncertain. We suggest to evaluate patients who have only some and not all of the core DOORS characteristics already for a *TBC1D24* mutation as only this way we will learn what the phenotype of DOORS syndrome can be, and study further clinical and basal aspects [Hennekam, 2007].

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REFERENCES

Azaiez H, Booth KT, Bu F, Huygen P, Shibata SB, Shearer AE, Kolbe D, Meyer N, Black-Ziegelbein EA, Smith RJ. 2014. TBC1D24 mutation causes autosomal-dominant nonsyndromic hearing loss. Hum Mutat 35: 819–823.

Campeau PM, Kasperaviciute D, Lu JT, Burrage LC, Kim C, Hori M, Powell BR, Stewart F, Felix TM, van den Ende J, Wisniewska M, Kayserili H, Rump P, Nampoothiri S, Aftimos S, Mey A, Nair LD, Begleiter ML, De Bie I, Meenakshi G, Murray ML, Repetto GM, Golabi M, Blair E, Male A, Giuliano F, Kariminejad A, Newman WG, Bhaskar SS, Dickerson JE, Kerr B, Banka S, Giltay JC, Wieczorek D, Tostevin A, Wiszniewska J, Cheung SW, Hennekam RC, Gibbs RA, Lee BH, Sisodiya SM. 2014. The genetic basis of DOORS syndrome: An exome-sequencing study. Lancet Neurol 13:44–58.

Cantwell RJ. 1975. Congenital sensori-neural deafness associated with onycho-osteo dystrophy and mental retardation (D.O.O.R. syndrome). Humangenetik 26:261–265.

Castori M, Valiante M, Pascolini G, Leuzzi V, Pizzuti A, Grammatico P. 2013. Clinical and genetic study of two patients with Zimmermann-Laband syndrome and literature review. Eur J Med Genet 56:570–576.

Corbett MA, Bahlo M, Jolly L, Afawi Z, Gardner AE, Oliver KL, Tan S, Coffey A, Mulley JC, Dibbens LM, Simri W, Shalata A, Kivity S, Jackson GD, Berkovic SF, Gecz J. 2010. A focal epilepsy and intellectual disability syndrome is due to a mutation in TBC1D24. Am J Hum Genet 87:371–375.

Eronen M, Somer M, Gustafsson B, Holmberg C, Fraser FC, Preus M. 1985. New syndrome: A digito-reno-cerebral syndrome. Am J Med Genet 22:281–285.

Falace A, Filipello F, La Padula V, Vanni N, Madia F, De Pietri Tonelli D, de Falco FA, Striano P, Dagna Bricarelli F, Minetti C, Benfenati F, Fassio A, Zara F. 2010. TBC1D24, an ARF6-interacting protein, is mutated in familial infantile myoclonic epilepsy. Am J Hum Genet 87:365–370.

Feinmesser M, Zelig S. 1961. Congenital deafness associated with onychodystrophy. Arch Otolaryngol 74:507–508.

Goodman RM, Lockareff S, Gwinup G. 1969. Hereditary congenital deafness with onychodystrophy. Arch Otolaryngol 90:474–477.

Guven A, Tolun A. 2013. TBC1D24 truncating mutation resulting in severe neurodegeneration. J Med Genet 50:199–202.

Hennekam RC. 2007. What to call a syndrome. Am J Med Genet A 143A: 1021–1024.

Jacquinet A, Gerard M, Gabbett MT, Rausin L, Misson JP, Menten B, Mortier G, Van Maldergem L, Verloes A, Debray FG. 2010. Temple-Baraitser syndrome: A rare and possibly unrecognized condition. Am J Med Genet A 152A:2322–2326.

James AW, Miranda SG, Culver K, Hall BD, Golabi M. 2007. DOOR syndrome: Clinical report, literature review and discussion of natural history. Am J Med Genet A 143A:2821–2831.

Le Merrer M, David A, Goutieres F, Briard ML. 1992. Digito-reno-cerebral syndrome: Confirmation of Eronen syndrome. Clin Genet 42:196–198.

Milh M, Falace A, Villeneuve N, Vanni N, Cacciagli P, Assereto S, Nabbout R, Benfenati F, Zara F, Chabrol B, Villard L, Fassio A. 2013. Novel compound heterozygous mutations in TBC1D24 cause familial malignant migrating partial seizures of infancy. Hum Mutat 34:869–872.

Qazi QH, Nangia BS. 1984. Abnormal distal phalanges and nails, deafness, mental retardation, and seizure disorder: A new familial syndrome. The Journal of Pediatrics 104: 391–394.

Rehman AU, Santos-Cortez RL, Morell RJ, Drummond MC, Ito T, Lee K, Khan AA, Basra MA, Wasif N, Ayub M, Ali RA, Raza SI. University of Washington Center for Mendelian G. Nickerson DA, Shendure J, Bamshad M, Riazuddin S, Billington N, Khan SN, Friedman PL, Griffith AJ, Ahmad W, Riazuddin S, Leal SM, Friedman TB. 2014. Mutations in TBC1D24, a gene associated with epilepsy, also cause nonsyndromic deafness DFNB86. Am J Hum Genet 94:144–152.

Santen GW, Aten E, Vulto-van Silfhout AT, Pottinger C, van Bon BW, van Minderhout IJ, Snowdowne R, van der Lans CA, Boogaard M, Linssen MM, Vijfhuizen L, van der Wielen MJ, Vollebregt MJ, Coffin-Siris C, Breuning MH, Kriek M, van Haeringen A, den Dunnen JT, Hoischen A, Clayton-Smith J, de Vries BB, Hennekam RC, van Belzen MJ. 2013. Coffin-Siris syndrome and the BAF complex: Genotype-phenotype study in 63 patients. Hum Mutat 34:1519–1528.

Sousa SB, Abdul-Rahman OA, Bottani A, Cormier-Daire V, Fryer A, Gillessen-Kaesbach G, Horn D, Josifova D, Kuechler A, Lees M, MacDermot K, Magee A, Morice-Picard F, Rosser E, Sarkar A, Shannon N, Stolte-Dijkstra I, Verloes A, Wakeling E, Wilson L, Hennekam RC. 2009. Nicolaides-Baraitser syndrome: Delineation of the phenotype. Am J Med Genet A 149A:1628–1640.

Tsurusaki Y, Okamoto N, Ohashi H, Kosho T, Imai Y, Hibi-Ko Y, Kaname T, Naritomi K, Kawame H, Wakui K, Fukushima Y, Homma T, Kato M, Hiraki Y, Yamagata T, Yano S, Mizuno S, Sakazume S, Ishii T, Nagai T, Shiina M, Ogata K, Ohta T, Niikawa N, Miyatake S, Okada I, Mizuguchi T, Doi H, Saitsu H, Miyake N, Matsumoto N. 2012. Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. Nat Genet 44:376–378.

Tsurusaki Y, Okamoto N, Ohashi H, Mizuno S, Matsumoto N, Makita Y, Fukuda M, Isidor B, Perrier J, Aggarwal S, Dalal AB, Al-Kindy A, Liebelt J, Mowat D, Nakashima M, Saitsu H, Miyake N, Matsumoto N. 2014. Coffin-Siris syndrome is a SWI/SNF complex disorder. Clin Genet 85:548–554.

Van Houdt JK, Nowakowska BA, Sousa SB, van Schaik BD, Seuntjens E, Avonce N, Sifrim A, Abdul-Rahman OA, van den Boogaard MJ, Bottani A, Castori M, Cormier-Daire V, Deardorff MA, Filges I, Fryer A, Fryns JP, Gana S, Garavelli L, Gillessen-Kaesbach G, Hall BD, Horn D, Huylebroeck D, Klapecki J, Krajewska-Walasek M, Kuechler A, Lines MA, Maas S, Macdermot KD, McKee S, Magee A, de Man SA, Moreau Y, Morice-Picard F, Obersztyn E, Pilch J, Rosser E, Shannon N, Stolte-Dijkstra I, Van Dijck P, Vilain C, Vogels A, Wakeling E, Wieczorek D, Wilson L, Zuffardi O, van Kampen AH, Devriendt K, Hennekam R, Vermeesch JR. 2012. Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nat Genet 44:445–449S1.

Walbaum R, Fontaine G, Lienhardt J, Piquet JJ. 1970. [Familial deafness with osteo-onychodysplasia]. J Genet Hum 18:101–108.

Winter RM. 1993. Eronen syndrome identical with DOOR syndrome? Clin Genet 43:167.

Zhang L, Hu L, Chai Y, Pang X, Yang T, Wu H. 2014. A dominant mutation in the stereocilia-expressing gene TBC1D24 is a probable cause for nonsyndromic hearing impairment. Hum Mutat 35:814–818.