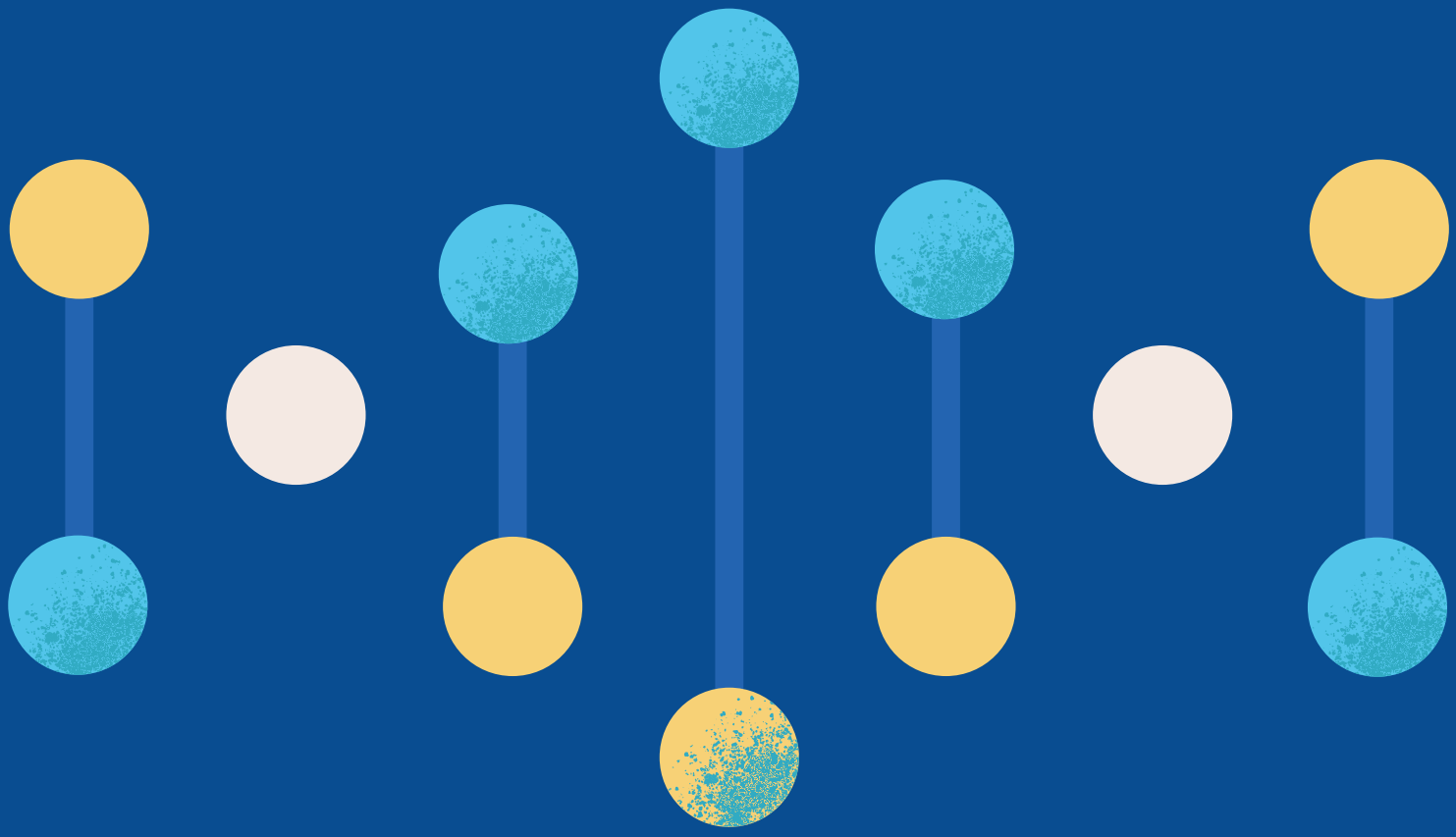
TGF-BR1 GENE COMPENDIUM

A Curated Reference for Thoracic Aortic Aneurysm and Dissection (TAAD)



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Notices
This compendium is provided as an educational resource.
Medical and genetic knowledge evolves rapidly, and users are advised to consult up-to-date literature and clinical guidance when interpreting genetic variants.

Curators have made every effort to ensure accuracy but bear no liability for clinical or diagnostic outcomes based on this document. All variant classifications and recommendations should be considered in context and with professional clinical judgment.

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1. Basic Information for TAAD Gene

1.1 Gene Identifiers

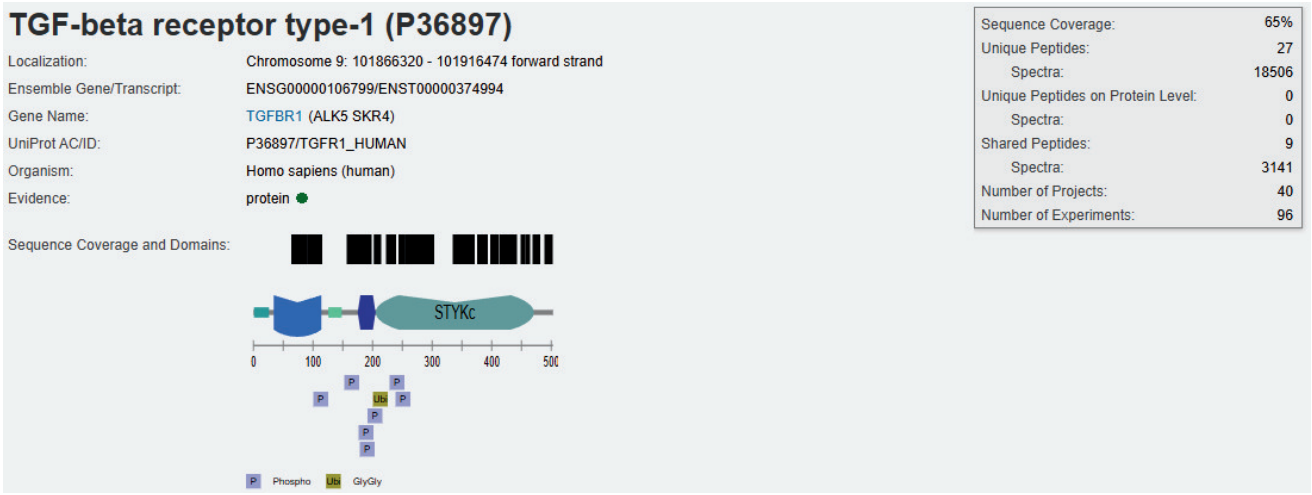
Gene MIM Number	190181
Phenotype MIM Number	609192 Loeys-Dietz syndrome1
Orphanet code	TAAD 91387 LDS 60030
Chromosomal Position (hg19/GRCh37)	Chr 9:101,867,395-101,916,474
Chromosomal Position (hg38/GRCh38)	Chr 9:99,105,113-99,154,192
Total Number of Amino Acids	503
MANE Transcript	NM_04612.4
Are there pseudogenes?	No
Other pitfalls (e.g. retrocopy insertion polymorphisms)?	Variation in Ala repetition in exon 1 may cause technical issues

1.2 Other phenotypes associated with TGF-βR1 with no or lower prevalence of TAAD

TGF-βR1 variants in extracellular domain leading to haplo-insufficiency are associated with Multiple self-healing squamous epithelioma (OMIM 132800).

2. Protein Structure Overview

2.1. Scheme with important domains within the protein



Source: https://www.proteomicsdb.org/proteomicsdb/#protein/proteinDetails/55234/summary

Table: Overview of TGF-βR1 protein with Amino Acid (AA) boundaries

Domain	From AA	To AA
Signal peptide	1	29
Extracellular	30	126
Trans-membrane	127	147
Non-specified	148	174
GS domain	175	204
Serine-Threonine Protein kinase	205	495

Source: www.umd.be/TGFBR1

2.2 List of Exon Boundaries (at cDNA Level)

MANE NM: 004612.4	Nucleotide Range	Amino Acid Range
5'UTR / Exon 1	c.-93 to 97	1 to 33
Exon 2	c.98 to 343	33 to 115

Exon 3	c.344 to 574	115 to 192
Exon 4	c.575 to 805	192 to 269
Exon 5	c.806 to 973	269 to 325
Exon 6	c.974 to 1130	325 to 377
Exon 7	c.1131 to 1255	377 to 419
Exon 8	c.1256 to 1386	419 to 462
Exon 9-3'UTR	c.1387 to *4887	463 to 503

Source: Alamut

2.3. Mutational hotspot or recurrent/founder variants

cDNA annotation	**Protein annotation**	**Number of submissions in ClinVar**
c.680AAG[1]	p.Glu228del	5
c.722C>T	p.Ser241Leu	5 VarID 213896
c.934G>A	p.Gly312Ser	7 VarID 12524
c.1459C>T	p.Arg487Trp	7 VarID 213882
c.1460G>A	p.Arg487Gln	9 VarID 12525

2.4. Major domain(s) that involves more pathogenic variants

Most variants located on the Serine-Threonine Protein kinase domain (aa205- aa495) (Loeys et al. 2006).

Variants also described on the GS-domain: aa175- aa204 (Loeys et al. 2005).

No described variants in the signal peptide, extracellular or transmembrane domain associated with Loeys-Dietz syndrome (LDS) or TAAD. PTC variants in those domains are associated with Multiple self-healing squamous epithelioma (MSSE).

PTC variants in serine-threonine kinase domain should be checked for escape of NMD. Current literature suggests that only PTC variants that escape NMD cause aneurysmal phenotype (but needs further confirmation) (Goudie et al. 2011).

> Clinvar database: ClinVar - TGFBR1
> GnomAD database: gnomAD.v4.1 TGFBR1
> Mutscore: MutScore TGFBR1

3. Mode of Inheritance

Loeys-Dietz syndrome (LDS) is inherited in an autosomal dominant manner. Wide intrafamilial phenotypic variability has been documented. Rare examples of non-penetrance in LDS have been documented. In one case, this was related to somatic mosaicism (Baban et al, 2018).

4. Constraint Matrices and Mutational Subtypes

pLI score	pLI=0,96 o/e= 0,35 (0.24-0.51)	gnomad v.4.1
Missense constraint	Z=3,65; o/e=0.6 (0.55-0.66)	gnomad v.4.1
Haplo-insufficiency (nonsense or out of frame splice)	No*	Goudie et al, 2011
Full gene deletion	No**	Redon et al, 2006
Multi-exon deletion	Yes***	Bichat lab
Full gene duplication	No****	Breckpot et al, 2010
Multi-exon duplication	No	

*PTC variants in serine-threonine kinase domain that escape NMD have been described in aneurysm phenotypes:

1. Fortugno et al 2023 described 2 different frameshift variations in exon 8 (escaping NMD).
2. In Bichat's laboratory, out of 104 MFS/TAAD probands with pathogenic variations in TGFBR1 gene, the following variations were identified:
   - Gln250* (ex4) in a proband with familial TAA (3 symptomatic carriers).
   - Leu406* (ex7) in a proband with familial TAA and some skeletal features, 9 carriers in the family (pedigree with 3 generations, 1 case of incomplete penetrance and 1 asymptomatic child).
   - Tyr476* (ex9) in a proband with TAA and mitral valve surgery (no family data).
   - c.805+1G>C (ex4) in a proband with familial TAAD (2 symptomatic carriers).

** the individual reported lacked aortic involvement (Redon et al. 2006).
*** a heterozygous deletion of exon 4 (predicted in frame) in a family with TAA, systemic features including bifid uvula (3 symptomatic carriers) was identified.
**** the individual reported lacked aortic involvement. (Breckpot et al. 2010).

5. Mode of Action of the Pathogenic Variants

Gain-of-function	No	—
Haploinsufficiency (as defined by allele that undergoes (or predicted to undergo NMD) or full gene deletion	No*	Lindsay and Dietz, 2011
Dominant-negative	Unknown**	Lindsay and Dietz, 2011
Loss-of-function (not driven by haplo-insufficiency)	Unknown*/**	Lindsay and Dietz, 2011

* the current body of evidence suggests that there must be some residual mutant protein expression necessary in order to cause an aneurysmal phenotype but that the precise mechanism of action remains under investigation.
** if mutant receptor is expressed by itself it demonstrates loss-of-function, but at the cellular membrane where it functions in a complex, the effect is more complex.

6. Additional Supportive Evidence for Pathogenicity

6.1. Clinical evidence

No features are specific for TGF-βR1.

6.2. Functional evidence

Currently, no functional tests are available that could help for missense variants interpretation in TGF-βR1. In the case of a nonsense mutation, efforts to examine nonsense-mediated decay (NMD) should be performed to demonstrate NMD escape before concluding on pathogenicity.

7. Mutation Database or Expert

7.1. Gene-specific mutation database

- UMD: UMD-TGFBR1
- LOVD: https://databases.lovd.nl/shared/genes/TGFBR1
- HGMD: https://www.hgmd.cf.ac.uk/ac/gene.php?gene=TGFBR1
- Clinvar: TGFBR1-ClinVar

7.2 Uniprot 3D protein structure

The UniProt database provides access to the 3D structure and functional annotation of TGF-βR1: https://www.uniprot.org/uniprotkb/P36897/entry#structure

7.3 Lab "expert" for this gene that can be contacted for advice on specific cases

For questions about specific variants or complex TGF-βR1 cases, expert consultation is available at

Center for Medical Genetics, University Hospital Antwerp, Belgium

- Bart.loeys@uza.be

Genetics Department, Bichat hospital, Paris, France

- pauline.arnaud@aphp.fr

8. Phenotype Characteristics

8.1. Estimated prevalence of the disease

The exact prevalence of LDS is unknown but estimated to be 1:50,000. More than 1,000 families with LDS have been described in the literature.

8.2. Youngest age of onset of aortic aneurysm

Neonatal (Yetman et al, 2007).

8.3. Youngest age of onset of aortic dissection

Three months (Williams et al, 2008).

8.4. genotype-phenotype correlation validated for this gene

A correlation was demonstrated between the severity of craniofacial involvement and the age at the first cardiovascular event, that is, surgery, dissection or death (Loeys et al, 2005; Jondeau et al, 2016).

Non-syndromic and syndromic presentations of aortic aneurysm/dissection have been associated with the identical TGF-βR1 variants (personal observation, Loeys and Dietz)
In Regalado et al 2022, Arg487 substitutions confer a higher risk for aortic events than other variants. So far, this does not change the clinical management of individuals with this specific variant.

8.5. Is there any consensus on management of the disease?

Yes, not specific for TGF-βR1, but for LDS in general (Mac Carrick et al, 2014). Also consult general TAAD guidelines (Isselbacher et al 2022, Mazzolai et al, 2024, Morris et al, 2024).

References

- Baban A, Magliozzi M, Loeys B, Adorisio R, Alesi V, Secinaro A, Corica B, Vricella L, Dietz HC, Drago F, Novelli A, Amodeo A. First evidence of maternally inherited mosaicism in TGFBR1 and subtle primary myocardial changes in Loeys-Dietz syndrome: a case report. BMC Med Genet. 2018 Sep 15;19(1):170. doi: 10.1186/s12881-018-0661-2. PMID: 30219046; PMCID: PMC6139163.
- Breckpot J, Budts W, De Zegher F, Vermeesch JR, Devriendt K. Duplication of the TGFBR1 gene causes features of Loeys-Dietz syndrome. Eur J Med Genet. 2010 Nov-Dec;53(6):408-10. doi: 10.1016/j.ejmg.2010.08.004. Epub 2010 Sep 20. PMID: 20813212.
- Camerota L, Ritelli M, Wischmeijer A, Majore S, Cinquina V, Fortugno P, et al., Genotypic categorization of Loeys-Dietz syndrome based on 24 novel families and literature data, Genes (Basel) 10 (10) (2019), doi: 10.3390/genes10100764. Epub 2019/10/02. (PubMed PMID: 31569402; PubMed Central PMCID: PMCPMC6826414)
- Erkula G, Sponseller PD, Paulsen LC, Oswald GL, Loeys BL, Dietz HC. Musculoskeletal findings of Loeys-Dietz syndrome. J Bone Joint Surg Am. 2010 Aug 4;92(9):1876-83. doi: 10.2106/JBJS.I.01140. PMID: 20686062.
- Felgentreff K, Siepe M, Kotthoff S, von Kodolitsch Y, Schachtrup K, Notarangelo LD, Walter JE, Ehl S. Severe eczema and Hyper-IgE in Loeys-Dietz-syndrome - contribution to new findings of immune dysregulation in connective tissue disorders. Clin Immunol. 2014 Jan;150(1):43-50. doi: 10.1016/j.clim.2013.11.008. Epub 2013 Nov 19. PMID: 24333532.
- Fortugno P, Monetta R, Cinquina V, Rigon C, Boaretto F, De Luca C, Zoppi N, Di Leandro L, De Domenico E, Di Daniele A, Ippoliti R, Angelucci F, Di Cesare E, De Paulis R, Salviati L, Colombi M, Brancati F, Ritelli M. Truncating variants in the penultimate exon of TGFBR1 escaping nonsense-mediated mRNA decay cause Loeys-Dietz syndrome. Eur J Hum Genet. 2023 May;31(5):596-601. doi: 10.1038/s41431-022-01279-4. Epub 2023 Jan 4. PMID: 36599937; PMCID: PMC10172188.
- Frischmeyer-Guerrerio PA, Guerrerio AL, Oswald G, Chichester K, Myers L, Halushka MK, Oliva-Hemker M, Wood RA, Dietz HC. TGFβ receptor mutations impose a strong predisposition for human allergic disease. Sci Transl Med. 2013 Jul 24;5(195):195ra94. doi: 10.1126/scitranslmed.3006448. PMID: 23884466; PMCID: PMC3905327.
- Frise CJ, Pitcher A, Mackillop L. Loeys-Dietz syndrome and pregnancy: The first ten years. Int J Cardiol. 2017 Jan 1;226:21-25. doi: 10.1016/j.ijcard.2016.10.024. Epub 2016 Oct 11. PMID: 27780078.
- Goudie DR, D'Alessandro M, Merriman B, Lee H, Szeverényi I, Avery S, O'Connor BD, Nelson SF, Coats SE, Stewart A, Christie L, Pichert G, Friedel J, Hayes I, Burrows N, Whittaker S, Gerdes AM, Broesby-Olsen S, Ferguson-Smith MA, Verma C, Lunny DP, Reversade B, Lane EB. Multiple self-healing squamous epithelioma is caused by a disease-specific spectrum of mutations in TGFBR1. Nat Genet. 2011 Feb 27;43(4):365-9. doi: 10.1038/ng.780. PMID: 21358634.
- Gutman G, Baris HN, Hirsch R, Mandel D, Yaron Y, Lessing JB, Kuperminc MJ. Loeys-Dietz syndrome in pregnancy: a case description and report of a novel mutation. Fetal Diagn Ther. 2009;26(1):35-7. doi: 10.1159/000236357. Epub 2009 Oct 10. PMID: 19816028.
- Loeys BL, Chen J, Neptune ER, Judge DP, Podowski M, Holm T, Meyers J, Leitch CC, Katsanis N, Sharifi N, Xu FL, Myers LA, Spevak PJ, Cameron DE, De Backer J, Hellemans J, Chen Y, Davis EC, Webb CL, Kress W, Coucke P, Rifkin DB, De Paepe AM, Dietz HC. A syndrome of altered cardiovascular, craniofacial, neurocognitive and skeletal development caused by mutations in TGFBR1 or TGFBR2. Nat Genet. 2005 Mar;37(3):275-81. doi: 10.1038/ng1511. Epub 2005 Jan 30. PMID: 15731757.

- Isselbacher EM, Preventza O, Hamilton Black J 3rd, Augoustides JG, Beck AW, Bolen MA, Braverman AC, Bray BE, Brown-Zimmerman MM, Chen EP, Collins TJ, DeAnda A Jr, Fanola CL, Girardi LN, Hicks CW, Hui DS, Schuyler Jones W, Kalahasti V, Kim KM, Milewicz DM, Oderich GS, Ogbechie L, Promes SB, Gyang Ross E, Schermerhorn ML, Singleton Times S, Tseng EE, Wang GJ, Woo YJ; Peer Review Committee Members. 2022 ACC/AHA Guideline for the Diagnosis and Management of Aortic Disease: A Report of the American Heart Association/American College of Cardiology Joint Committee on Clinical Practice Guidelines. Circulation. 2022 Dec 13;146(24):e334-e482. doi: 10.1161/CIR.0000000000001106. Epub 2022 Nov 2. PMID: 36322642; PMCID: PMC9876736.
- Jondeau G, Ropers J, Regalado E, Braverman A, Evangelista A, Teixedo G, De Backer J, Muiño-Mosquera L, Naudion S, Zordan C, Morisaki T, Morisaki H, Von Kodolitsch Y, Dupuis-Girod S, Morris SA, Jeremy R, Odent S, Adès LC, Bakshi M, Holman K, LeMaire S, Milleron O, Langeois M, Spentchian M, Aubart M, Boileau C, Pyeritz R, Milewicz DM; Montalcino Aortic Consortium. International Registry of Patients Carrying TGFBR1 or TGFBR2 Mutations: Results of the MAC (Montalcino Aortic Consortium). Circ Cardiovasc Genet. 2016 Dec;9(6):548-558. doi: 10.1161/CIRCGENETICS.116.001485. Epub 2016 Nov 21. PMID: 27879313; PMCID: PMC5177493.
- Lindsay ME, Dietz HC. Lessons on the pathogenesis of aneurysm from heritable conditions. Nature. 2011 May 19;473(7347):308-16. doi: 10.1038/nature10145. PMID: 21593863; PMCID: PMC3622871.
- Loeys BL, Schwarze U, Holm T, Callewaert BL, Thomas GH, Pannu H, De Backer JF, Oswald GL, Symoens S, Manouvrier S, Roberts AE, Faravelli F, Greco MA, Pyeritz RE, Milewicz DM, Coucke PJ, Cameron DE, Braverman AC, Byers PH, De Paepe AM, Dietz HC. Aneurysm syndromes caused by mutations in the TGF-beta receptor. N Engl J Med. 2006 Aug 24;355(8):788-98. doi: 10.1056/NEJMoa055695. PMID: 16928994.
- MacCarrick G, Black JH 3rd, Bowdin S, El-Hamamsy I, Frischmeyer-Guerrerio PA, Guerrerio AL, Sponseller PD, Loeys B, Dietz HC 3rd. Loeys-Dietz syndrome: a primer for diagnosis and management. Genet Med. 2014 Aug;16(8):576-87. doi: 10.1038/gim.2014.11. Epub 2014 Feb 27. PMID: 24577266; PMCID: PMC4131122.
- Maleszewski JJ, Miller DV, Lu J, Dietz HC, Halushka MK. Histopathologic findings in ascending aortas from individuals with Loeys-Dietz syndrome (LDS). Am J Surg Pathol. 2009 Feb;33(2):194-201. doi: 10.1097/PAS.0b013e31817f3661. PMID: 18852674.
- Mazzolai L, Teixido-Tura G, Lanzi S, Boc V, Bossone E, Brodmann M, Bura-Rivière A, De Backer J, Deglise S, Della Corte A, Heiss C, Kałużna-Oleksy M, Kurpas D, McEniery CM, Mirault T, Pasquet AA, Pitcher A, Schaubroeck HAI, Schlager O, Sirnes PA, Sprynger MG, Stabile E, Steinbach F, Thielmann M, van Kimmenade RRJ, Venermo M, Rodriguez-Palomares JF; ESC Scientific Document Group. 2024 ESC Guidelines for the management of peripheral arterial and aortic diseases. Eur Heart J. 2024 Sep 29;45(36):3538-3700. doi: 10.1093/eurheartj/ehae179. PMID: 39210722.
- Morris SA, Flyer JN, Yetman AT, Quezada E, Cappella ES, Dietz HC, Milewicz DM, Ouzounian M, Rigelsky CM, Tierney S, Lacro RV; American Heart Association Council on Lifelong Congenital Heart Disease and Heart Health in the Young (Young Hearts); Council on Cardiovascular and Stroke Nursing; Council on Peripheral Vascular Disease; Council on Cardiopulmonary, Critical Care, Perioperative and Resuscitation; and Council on Cardiovascular Surgery and Anesthesia. Cardiovascular Management of Aortopathy in Children: A Scientific Statement From the American Heart Association. Circulation. 2024 Sep 10;150(11):e228-e254. doi: 10.1161/CIR.0000000000001265. Epub 2024 Aug 12. PMID: 39129620.

- Mühlstädt K, De Backer J, von Kodolitsch Y, Kutsche K, Muiño Mosquera L, Brickwedel J, Girdauskas E, Mir TS, Mahlmann A, Tsilimparis N, Staebler A, Schoof L, Seidel H, Berger J, Bernhardt AM, Blankenberg S, Kölbel T, Detter C, Szöcs K, Kaemmerer H. Case-matched Comparison of Cardiovascular Outcome in Loeys-Dietz Syndrome versus Marfan Syndrome. J Clin Med. 2019 Nov 29;8(12):2079. doi: 10.3390/jcm8122079. PMID: 31795342; PMCID: PMC6947024.
- Gouda P, Kay R, Habib M, Aziz A, Aziza E, Welsh R. Clinical features and complications of Loeys-Dietz syndrome: A systematic review. Int J Cardiol. 2022 Sep 1;362:158-167. doi: 10.1016/j.ijcard.2022.05.065. Epub 2022 Jun 1. PMID: 35662564.
- Redon R, Baujat G, Sanlaville D, Le Merrer M, Vekemans M, Munnich A, Carter NP, Cormier-Daire V, Colleaux L. Interstitial 9q22.3 microdeletion: clinical and molecular characterisation of a newly recognised overgrowth syndrome. Eur J Hum Genet. 2006 Jun;14(6):759-67. doi: 10.1038/sj.ejhg.5201613. PMID: 16570072.
- Regalado ES, Morris SA, Braverman AC, Hostetler EM, De Backer J, Li R, Pyeritz RE, Yetman AT, Cervi E, Shalhub S, Jeremy R, LeMaire S, Ouzounian M, Evangelista A, Boileau C, Jondeau G, Milewicz DM. Comparative Risks of Initial Aortic Events Associated With Genetic Thoracic Aortic Disease. J Am Coll Cardiol. 2022 Aug 30;80(9):857-869. doi: 10.1016/j.jacc.2022.05.054. PMID: 36007983.
- Teixidó-Tura G, Franken R, Galuppo V, Gutiérrez García-Moreno L, Borregan M, Mulder BJ, García-Dorado D, Evangelista A. Heterogeneity of aortic disease severity in patients with Loeys-Dietz syndrome. Heart. 2016 Apr;102(8):626-32. doi: 10.1136/heartjnl-2015-308535. Epub 2016 Feb 4. PMID: 26848186.
- Van Hemelrijk C, Renard M, Loeys B. The Loeys-Dietz syndrome: an update for the clinician. Curr Opin Cardiol. 2010 Nov;25(6):546-51. doi: 10.1097/HCO.0b013e32833f0220. PMID: 20838339.
- Wessels MW, Catsman-Berrevoets CE, Mancini GM, Breuning MH, Hoogeboom JJ, Stroink H, Frohn-Mulder I, Coucke PJ, Paepe AD, Niermeijer MF, Willems PJ. Three new families with arterial tortuosity syndrome. Am J Med Genet A. 2004 Dec 1;131(2):134-43. doi: 10.1002/ajmg.a.30272. PMID: 15529317.
- Williams JA, Loeys BL, Nwakanma LU, Dietz HC, Spevak PJ, Patel ND, François K, DeBacker J, Gott VL, Vricella LA, Cameron DE. Early surgical experience with Loeys-Dietz: a new syndrome of aggressive thoracic aortic aneurysm disease. Ann Thorac Surg. 2007 Feb;83(2):S757-63; discussion S785-90. doi: 10.1016/j.athoracsur.2006.10.091. PMID: 17257922.
- Yetman AT, Beroukhim RS, Ivy DD, Manchester D. Importance of the clinical recognition of Loeys-Dietz syndrome in the neonatal period. Pediatrics. 2007 May;119(5):e1199-202. doi: 10.1542/peds.2006-2886. PMID: 17470566; PMCID: PMC3131201.

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VASCERN, the European Reference Network on Rare Multisystemic Vascular Diseases, is dedicated to gathering the best expertise in Europe in order to provide accessible cross-border healthcare to patients with rare vascular diseases (an estimated 1.3 million affected). These include arterial diseases (affecting the aorta to small arteries), arterio-venous anomalies, vascular malformations, and lymphatic diseases.

VASCERN currently comprises 48 expert teams from 39 highly specialised multidisciplinary HCPs coming from 19 EU Member States, as well as various European Patient Organisations, and is coordinated in Paris, France.

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