DESSH Publications *(as of 4/2026)*

2002	Xu G.M., Arnaout M.A. (**2002**). WAC, a novel WW Domain-Containing Adapter with a Coiled-Coil Region, is colocalized with splicing factor SC35. *Genomics 79*:87–94.
2004	Yatsenko S.A., Yatsenko A.N., Szigeti K., Craigen W.J., Stankiewicz P., Cheung S.W., et al. (**2004**). Interstitial deletion of 10p and atrial septal defect in DiGeorge 2 syndrome. *Clin Genet*, 66:128-136.
2008	Shadadpuri R., de Vries B., Pfundt R., de Leeuw N., & Reardon W. (**2008**). Pseudoarthrosis of the clavicle and copper beaten skull associated with chromosome 10p11.21p12.1 microdeletion. *Amer J Med Genet, Part A,* 146:233-237. Https://doi.org/10.1002/ajmg.a.32088
2011	Totsukawa G., Kaneko Y., Uchiyama K., Toh H., Tamura K., Kondo H. **(2011)**. VCIP135 deubitiquinase and its binding protein, WAC, in p97ATPase-mediated membrane fusion. *EMBO Journal*, 2011; 30(17). Https://doi.org/10.1038/emboj.2011.260
2011	Wentzel C., Rajcan-Separovic E., Ruivenkamp C.A.L., Chantot-Bastaraud S., Metay C., Andrieux J., Annerén G., Gijsbers A.C.J., Druart L., Hyon C., Portnoi M.F., Stattin E.L., Vincent-Delorme C., Kant S.G., Steinraths M., Marlin S., Giurgea I., & Thuresson A.C. (**2011**). Genomic and clinical characteristics of six patients with partially overlapping interstitial deletions at 10p12p11. *European Journal of Human Genetics*, 19:959-964. Https://doi.org/10.1038/ejhg.2011.71
2011	Zhang F., Yu X. (**2011**). WAC, a functional partner of RNF20/40, regulates histone H2B ubiquitination and gene transcription. *Mol Cell* 41(2011):384-397.
2012	Joachim J., Wirth M., McKnight N.C., Tooze S.A. **(2012)**. Coiling up with SCOC and WAC: Two new regulators of starvation-induced autophagy. *Autophagy*, 2012; 8(9). Https://doi.org/10.4161/auto.21043
2012	McKnight N.C., Jefferies H.B.J., Alemu E.A., Saunders R.E., Howell M., Johansen T., Tooze S.A. **(2012).** Genome-wide SiRNA screen reveals amino acid starvation-autophagy requires SCOC and WAC. EMBO J. 31:1931–1946.
2012	Okamoto N., Hayashi S., Masui A., Kosaki R., Oguri I., Hasegawa T., Imoto I., Makita Y., Hata A., Moriyama K., & Inazawa J. (**2012**). Deletion at chromosome 10p11.23-p12.1 defines characteristic phenotypes with marked midface retrusion. *Journal of Human Genetics*, 57:191-196. Https://doi.org/10.1038/jhg.2011.154
2014	Hamdan F.F., Srour M., Capo-Chichi J.-M., Daoud H., Nassif C., Patry L., Massicotte C., Ambalavanan A., Spiegelman D., Diallo O., Henrion E., Dionne-Laporte A., Fougerat A., Pshezhetsky A.V., Venkateswaran S., Rouleau G.A., Michaud J.L. (**2014**). De novo mutations in moderate or severe intellectual disability. *PLoS Genet.* 10:31004772, 2014. Electronic article.
2014	Imoto H.J., Arnold G., Schneck F.X., Rajkovic A., and Yatsenko S.A. (**2014**). Interstitial 10p11.23-p12.1 microdeletions associated with developmental delay, craniofacial abnormalities, and cryptorchidism. *American Journal of Medical Genetics Part A*, vol. 164, no. 10, pp. 2623–2626.
2014	Mroczkowski H.J., Arnold G., Schneck F.X., Rajkovic A., & Yatsenko S. A. (**2014**). Interstitial 10p11. 23–p12.1 microdeletions associated with developmental delay, craniofacial abnormalities, and cryptorchidism. *Am J Med Genet A.* 164:2623–2626. https://doi:10.1002/ajmg.a.36627
2015	DeSanto C., D'Aco K., Araujo G.C., Shannon N., DDD Study, Vernon H., Rahrig A., Monaghan K.G., Niu Z., Vitazka P., Dodd J., Tang S., Manwaring L., Martir-Negron A.,

DESSH Publications *(as of 4/2026)*

	Schnur R.E., Juusola J., Schroeder A., Pan V., Helbig K.L., Friedman B., Shinawi M. (**2015**). *WAC* loss-of-function mutations cause a recognisable syndrome characterised by dysmorphic features, developmental delay and hypotonia and recapitulate 10p11.23 microdeletion syndrome. *Journal of Medical Genetics,* 52:754– 761. Https://doi.org/10.1136/jmedgenet-2015-103069
2015	Joachim J., Jefferies H.B.J., Razi M., *et al.* **(2015)**. Activation of ULK Kinase and autophagy by GABARAP trafficking from the centrosome is regulated by WAC and GM130. *Mol. Cell.* 2015; 60(6). Https://doi.org/10.1016/j.molcel.2015.11.018
2015	Sanders S.J., He X., Willsey A.M., Ercan-Sencicek A.G., Samocha K.E., Cicek A.E., Murtha M.T., Bal V.H., Bishop S.L., Dong S., et al. (**2015**). Insights into Autism Spectrum Disorder genomic architecture and biology from 71 risk loci. *Neuron* 87:1215-1233.
2015	Sosoi S., Streata I., Tudorache S., et al. (**2015**). Prenatal and postnatal findings in a 10.6 Mb interstitial deletion at 10p11.22–p12.31. *J Hum Genet.* 60:183–185. doi:10.1038/jhg.2015.4
2016	Abdelhedi F., El Khattabi L., Essid N., Viot G., Letessier D., Lebbar A., Dupont J.M. (**2016**). A *de novo* 10p11.23– p12.1 deletion recapitulates the phenotype observed in WAC mutations and strengthens the role of WAC in intellectual disability and behavior disorders. *Am J Med Genet A*. 170:1912–1917. https://doi:10.1002/ajmg.a.37686
2016	David-Morrison G., Xu Z., Rui Y., Chamg W.-L., Jaiswal M., Yamamoto S., Xiong B., Zhang K., Sandoval H., Duraine L., et al**. (2016).** WAC regulates mTOR activity by acting as an adaptor for the TTT and Pontin/Reptin complexes. *Developmental Cell* 36, 139–151.
2016	Lugtenberg D., Reijnders M.R.F., Fenckova M., Bijlsma E.K., Bernier R., van Bon B.W.M., Smeets E., Vulto-van Silfhout A.T., Bosch D., Eichler E.E., Mefford H.C., Carvill G.L., Bongers E.M.H.F., Schuurs- Hoeijmakers J.H.M., Ruivenkamp C.A., Santen G.W.E., van den Maagdenberg A.M.J.M., Peeters-Scholte C.M.P.C.D., Kuenen S., Verstreken P., Pfundt R., Yntema H.G., de Vries P.F., Veltman J.A., ... Vissers L.E.L.M. (**2016**). De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in *Drosophila*. *European Journal of Human Genetics*, 24:1145-1153. Https://doi.org/10.1038/ejhg.2015.282
2016	Rahrig D., Reijnders M.R.F., Fenckova M. et al. (**2016**). *De novo* loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in *Drosophila*. *European Journal of Human Genetics* 24(8):1145–1153.
2017	Varvagiannis K., de Vries B.B.A., Vissers L.E.L.M. (**2017**). WAC-related intellectual disabiity. In: M.P. Adam, H.H. Ardinger, R.A. Pagon, S.E. Wallace, L.J. Bean, K. Stephens, & A. Amemiya, eds. *GeneReviews*. Seattle, Washington, University of Washington.
2018	Qi F., Chen Q., Chen H., Yan H., Chen B., Xiang X., Liang C., Yi Q., Zhang M., Cheng H., et al. (**2018**). WAC promotes polo-like kinase 1 activation for timely mitotic entry. *Cell Rep. 24*:546–556.
2018	Uehara T., Ishige T., Hattori S., Yoshihashi H., Funato M., Yamaguchi Y., Takenouchi T., & Kosaki K. (**2018**). Three patients with DeSanto-Shinawi syndrome: Further phenotypic delineation. *Amer J Med Genet, Part A,* 176:1335-1340. Https://doi.org/10.1002/ajmg.a.38703

DESSH Publications *(as of 4/2026)*

2018	Vanegas S., Ramirez-Montanõ D., Candelo E., Shinawi M., & Pachajoa H. (**2018**). DeSanto-Shinawi syndrome: First case in South America. *Molecular Syndromology*, 9:154-158. Https://doi.org/10.1159/000488815
2018	Vissers L., de Vries B., Varvagiannis K. & Dingemans L. (**201**8). Human disease genes, website series: WAC. Human Disease Genes. https://humandiseasegenes.nl/wac/graph-and-chart/
2019	Zhang Y.J., Yao P.L., Zhou Y.F., Qiu T., Wang J., Wang X.H., Zhou S.Z., Wu B.B., & Wang Y. (**2019**). WAC gene pathogenic variation cause DeSanto-Shinawi syndrome with electrical status epilepticus during sleep. *Chinese Journal of Pediatrics*, 57:802-804. Https://doi.org/10.3760/cma.j.issn.0578-1310.2019.10.015
2020	Alsahlawi, Z., Jailani, M., Alaradi, H., & AlAbbad, A. (**2020**). A case of DeSanto-Shinawi syndrome in Bahrain with a novel mutation. *Case Reports in Pediatrics*, 2020, 1– 6. Https://doi.org/10.1155/2020/8820966
2020	Leonardi E., Bellini M., Aspromonte M.C., Polli R., Mercante A., Ciaccio C., Granocchio E., Bettella E., Donati I., Cainelli E., Boni S., Sartori S., Pantaleoni C., Boniver C. & Murgia A. (**2020**). A novel WAC loss of function mutation in an individual presenting with encephalopathy related to status epilepticus during sleep (ESES). *Genes*, 11:344. Https://doi.org/10.3390/genes11030344
2020	Orman A., Hakan N., Cagler A., Aydin M., Taskin E. (**2020**). Hypertrichosis in a newborn with deletion of the short arm of chromosome 10 (10p12.1). *Pediatric Oncall Journal*, 17(2):61-62. https://doi.org/10/7199/pede.oncall.2020.17
2021	Alawadhi, A., Morgan, A.T., Mucha, B.E., Scheffer, I.E., & Myers, K.A. (**2021**). Self-limited focal epilepsy and childhood apraxia of speech with WAC pathogenic variants. *European Journal of Paediatric Neurology*, 30:25– 28. 2021. Https://doi.org/10.1016/j.ejpn.2020.12.010
2021	Meng D., Guo K., Zhang D., Zhao C., Sun C., & Zhang F. (**2021**). Ring finger 20/ring finger 40/WW domain-containing adaptor with coiled- coil complex interacts with p53 to regulate gene transcription in DNA damage response. *Oncology Letters*, 21(6), 436. https://doi.org/10. 3892/ol.2021.12697
2021	Prentice P. and Gevers E. (**2021**)**.** Short stature due to a WAC mutation in Desanto-Shinawi Syndrome. (*abstract*). *Endocrine Abstracts,* 78:P54. (Poster presentation at British Society of Paediatric Endocrinology and Diabetes BSPED 2021). https://DOI:10.1530/endoabs.78.P54 .
2021	Takajo D., Katato G., Aggarwal S. (**2021**). Rapid progression of aortic stenosis in a 3-month-old infant with bicuspid aortic valve and DeSanto-Shinawi syndrome. *Ann Pediatr Cardiol*, 14(2):208-210. https://doi:10.4103/apc.APC_20_20
2022	Bolat H., Derin H., Ünsel-Bolat G. (**2022**). Phenotypic and brain imaging findings associated with a 10p proximal deletion including the *WAC* gene: Case report and literature review. *Cognitive and Behavioral Neurology* 35(3):221-226. https://DOI:10.1097/WNN.0000000000000309 .
2022	Branco J., Amorim M., Conde M. (**2022**). A novel variant of DeSanto-Shinawi syndrome with joint manifestations. *European Journal of Medical Genetics* 65(2022) 104534.
2022	Fu J.M., Satterstrom F.K., Peng M., Brand H., Collins R.L., Dong, S., Wamsley B., Klei L., Wang L., Hao S.P., et al. (**2022**). Rare coding variation provides insight into the genetic architecture and phenotypic context of autism. *Nat. Genet. 54*:1320–1331. *(On WAC and many other genes)*

DESSH Publications *(as of 4/2026)*

2022	Ho, S., Luk, H.-M., & Lo, I. F. M. **(2022**). Extending the phenotype of DeSanto-Shinawi syndrome: A case report and literature review. American Journal of Medical Genetics Part A, 188A:984–990. https://doi. org/10.1002/ajmg.a.62571
2022	Morales J. A., Valenzuela I., Cisco I., Cogné B., Isidor B., Matalon D. R., & Gomez-Ospina N. (**2022**). Clinical and molecular characterization of five new individuals with WAC-related intellectual disability: Evidence of pathogenicity for a novel splicing variant. *American Journal of Medical Genetics Part A*, 188A:1396–1406. https://doi.org/ 10.1002/ajmg.a.62648
2022	Quental R., Gonçalves D., Rodrigues E., Gonçalves E.S., Oliveira J., Freixo J. P., Leão M. **(2022).** Congenital heart defects associated with pathogenic variants in WAC gene: Expanding the phenotypic and genotypic spectrum of DeSanto-Shinawi Syndrome. *Am J Med Genet A*. 2022 Apr; 188(4):1311-1316. https://doi:10.1002/ajmg.a.62636.
2022	Stafford A.M., Pacheco-Vergara M., Uhl K.L., Jager T.E., Li X., Jeong J., Vogt D. (**2022**). A murine Wac model exhibits phenotypes relevant to DeSanto–Shinawi Syndrome. *bioRxiv* **2022**. doi: https://doi.org/10.1101/2022.01.24.477600
2022	Toledo-Gotor C., García-Muro C., García-Oguiza A., Poch-Olivé Ma. L., Ruiz-del Prado Ma. Y., Domínguez-Garrido E. (**2022**). Phenotypic comparison of patients affected with DeSanto-Shinawi syndrome: Point mutations in WAC gene versus a 10p12.1 microdeletion including WAC. *Mol Genet Genomic Med* 10(5):e1910. May 2022. https://doi.org/10.1002/mgg3.1910
2023	Anonymous. Data on Autism Detailed by Researchers at Michigan State University (Structure-Function of the Human WAC Protein in GABAergic Neurons: Towards an Understanding of Autosomal Dominant DeSanto-Shinawi Syndrome). (**2023**). *Mental Health Weekly Digest*, pg. 102. 2023.
2023	Nishikawa M., Matsuki T., Hamada N., Nakayama A., Ito H., Nagata K.-I. (**2023**). Expression analyses of WAC, a responsible gene for neurodevelopmental disorders, during mouse brain development. *Med Molec Morph 2023*. DOI https://doi.org/10.1007/s00795-023-00364-x
2023	Pasquali D., Torella A., Grandone A., Luongo C., Morleo M., Peduto C., di Fraia R., Selvaggio L.D., Allosso F., Accardo G., Zenobio M.T., Maitz S., Mariani M., Selicorni A., Banfi S., Nigro V. (**2023**). Patients with DeSanto-Shinawi syndrome: Further extension of phenotype from Italy. *Am J Med Genet* 2023; 191A:823-830. 2023. Https://DOI:10.1002/ajmg.a.63061
2023	Rudolph H.C., Stafford A.M., Hwang H.-E., Kim C.-H., Prokop J.W., Vogt D. (**2023**). Structure- Function of the Human WAC Protein in GABAergic Neurons: Towards an Understanding of Autosomal Dominant DeSanto–Shinawi Syndrome. *Biology* 2023, 12, 589. https://doi.org/10.3390/biology12040589
2024	Chen CP, Chen CY, Wu FT, Pan YT, Wu PS, Wang W. **(2024).** Prenatal diagnosis of a de novo 10p12.1p11.23 microdeletion encompassing the WAC gene in a fetus associated with bilateral hydronephrosis and right clubfoot on prenatal ultrasound. *Taiwan J Obstet Gynecol.* 2024 Jul;63(4):545-548. https://doi:10.1016/j.tjog.2024.05.008.
2024	Mail C., Yalcintepe S., Eker D., Gurkan H. **(2024).** The phenotypic spectrum of Desanto-Shinawi Syndrome: A comparative report of the first reported case in Turkey. *Genet Test Mol Biomarkers*. 2024 May; 28(5):213-217. https://doi:10.1089/gtmb.2023.0285.

DESSH Publications *(as of 4/2026)*

2024	Rahbeeni Z., Alwadani N., Al-Shehhi M., Faqeih E.A., Mohamed S. **(2024).** Report of DeSanto-Shinawi Syndrome in three boys with two novel variants in the WAC gene and expansion of the phenotype. *Cureus*. 2024 Oct 4;16(10):e70845. https://doi:10.7759/cureus.70845.
2024	Reynolds M., Weisenberg J., Shinawi M., Jensen R. **(2024)**. The DESSH Clinic: A new multidisciplinary clinic to address the complex needs of individuals with a rare genetic disorder. *Mo Med.* 2024 Jul-Aug;121(4):304-309. Www.msma.org/missouri-medicine
2024	Santoro G., Incoronato M., Spagnoli E. Gabbiato I., Contini S., Piovan M., Ferrari M., Lapucci C., Zuccarello D. **(2024).** An unclassified deletion involving the proximal short arm of Chromosome 10: A new syndrome? *Genes*, 2024, 15, 650. https://doi.org/10.3390/genes15060650
2025	Dwivedi A., Chauhan L., Kumar P., Nanda A., Jayakrishnan V.Y. **(2025).** Novel WAC gene variant identified in the first documented case of DeSanto-Shinawi Syndrome in India. *Mol Cell Pediatr* 2025 Nat 10, 12(1):7. https://doi:10.1186/s40348-025-00193-1.
2025	Lee K.H., Stafford A.M., Pacheco-Vergara M., Cichewicz K., Canales C.P., Seban N., Corea M., Rahbarian D., Bonekamp K.E., Gillie G.R., Cruz D.P., Gill A.M., Hwang H.E., Uhl K.L., Jager T.E., Shinawi M., Li X., Obenaus A., Crandall S., Jeong J., Nord A., Kim C.H., Vogt D.. Complimentary vertebrate *Wac* models exhibit phenotypes relevant to DeSanto-Shinawi Syndrome. *bioRxiv* [**Preprint**-not peer-reviewed yet]. 2025 Aug 28:2024.05.26.595966. https://doi:10.1101/2024.05.26.595966.
2025	Petroni S, Catena G, Iarossi G, Federici M, Zinzanella G, De Sanctis CM, Valente P, Buzzonetti L. **(2025).** Torpedo maculopathy in a patient with DeSanto-Shinawi syndrome. *Eur J Ophthalmol.* 2025 May;35(3):NP7-NP9. https://doi:10.1177/11206721251313840.
2026	Cipri S., Cacchione A., Agolini E., Verrigni D., Novelli A., Pepi C., De Palma L., De Benedictis A., Carai A., Barresi S., Rossi S., Alaggio R., Colafati G.S., D'Orazio D., Boccuto L., Mastonuzzi A. **(2026).** First report of a child with a DeSanto-Shinawi Syndrome and a polymorphous low-grade neuroepithelial tumor of the young. *Am J Med Genet A* 2026 (online ahead of print). https://doi:10.1002/ajmg.a.70085.
2026	Okamoto N, Nishi E, Hasegawa Y, Hayashi S. **(2026).** A patient with intellectual disability, agenesis of corpus callosum, and congenital heart disease associated with chromosome 10p11.2 microdeletion. *Am J Med Genet A*. 2026 Feb 2. https://doi:10.1002/ajmga.70070. ***Epub ahead of print.***