

Hereditary Angioedema (HAE) References

- Agostoni, A., Aygoren-Pursun, E., Binkley, K.E., Blanch, A., Bork, K., Bouillet, L., ... Zingale, L. (2004). Hereditary and acquired angioedema: problems and progress: proceedings of the third C1 esterase inhibitor deficiency workshop and beyond. *Journal of Allergy and Clinical Immunology*, 114 (3 Suppl), S51-131. <https://doi.org/10.1016/j.jaci.2004.06.047>
- Agostoni, A. & Cicardi, M., (1992). Hereditary and acquired C1-inhibitor deficiency: biological and clinical characteristics in 235 patients. *Medicine*, 71(4), 206-215.
https://journals.lww.com/md-journal/Citation/1992/07000/Heredity_and_Acquired_C1_Inhibitor_Deficiency_3.aspx
- Agostoni, A., Cicardi, M., Cugno, M., Zingale, L. C., Gioffre, D., & Nussberger, J. (1999). Angioedema due to angiotensin-converting enzyme inhibitors. *Immunopharmacology*, 44 (1-2), 21–25. [https://doi.org/10.1016/s0162-3109\(99\)00107-1](https://doi.org/10.1016/s0162-3109(99)00107-1)
- Aygören-Pürsün, E., Bygum, A., Grivcheva-Panovska, V., Magerl, M., Graff, J., Steiner, U. C., ... Cicardi, M. (2018). Oral Plasma Kallikrein Inhibitor for Prophylaxis in Hereditary Angioedema. *New England Journal of Medicine*, 379(4), 352-362.
<https://www.nejm.org/doi/full/10.1056/NEJMoa1716995>
- Baumgart, K. W., Britton, W. J., Kemp, A., French, M., & Robertson D. (1997). The spectrum of primary immunodeficiency disorders in Australia. *Journal of Allergy and Clinical Immunology*, 100(3), 415-423. [https://doi.org/10.1016/S0091-6749\(97\)70257-4](https://doi.org/10.1016/S0091-6749(97)70257-4)
- Bell, C. G., Kwan, E., Nolan, R. C., & Baumgart, K. W. (2008). First molecular confirmation of an Australian case of type III hereditary angioedema. *Pathology*, 40(1), 82-83.
https://journals.lww.com/pathologyrcpa/Citation/2008/40010/First_molecular_confirmation_of_an_Australian_case.15.aspx
- Bernstein, J. A., Levy, R., Wasserman, A., Bewtra, A., Hurewitz, D., Obtulowicz, K., ... Craig, T. J., (2008). Treatment of Acute Abdominal and Facial Attacks of Hereditary Angioedema (HAE) with Human C1-Esterase Inhibitor (C1-INH): Results of a Global, Multicenter, Randomized, Placebo-controlled, Phase II /III Study (I.M.P.A.C.T.1). *Journal of Allergy and Clinical Immunology*, 121(3), 795. <https://doi.org/10.1016/j.jaci.2008.01.052>
- Bonner, N., Abetz-Webb, L., Renault, L., Caballero, T., Longhurst, H., Maurer, M., Christiansen, S., & Zuraw, B. (2015). Icatibant Outcome Survey (IOS) International Executive Committee and the Hereditary Angioedema Association (HAEA) Medical Advisory Board. *Health Qual Life Outcomes*, 13, 92. <https://doi.org/10.1186/s12955-015-0292-7>

- Bork, K., Barnstedt, S., Koch, P., & Traupe, H. (2000). Hereditary angioedema with normal C1-inhibitor activity in women. *Lancet*, 356 (9225), 213-217. [https://doi.org/10.1016/S0140-6736\(00\)02483-1](https://doi.org/10.1016/S0140-6736(00)02483-1)
- Bork, K., Siedlecki, K., Bosch, S., Schopf, R.E., & Kreuz, W. (2000). Asphyxiation by laryngeal edema in patients with hereditary angioedema. *Mayo Clinical Procedures*, 75(4), 349-354. <https://doi.org/10.4065/75.4.349>
- Bork, K., & Barnstedt, S. (2001). Treatment of 193 episodes of laryngeal oedema with C1-INH concentrate in patients with hereditary angioedema. *Archives of Internal Medicine*, 161(5), 714-718. <https://doi.org/10.1001/archinte.161.5.714>
- Bork, K., Hardt, J., Schicketanz, K. H., & Ressel, N. (2003). Clinical Studies of Sudden Upper Airway Obstruction in Patients with Hereditary Angioedema Due to C1 Esterase Inhibitor Deficiency. *Archives of Internal Medicine*, 163(10), 1229–1235. <https://jamanetwork.com/journals/jamainternalmedicine/fullarticle/215579>
- Bork, K., Staubach, P., Eckardt, A. J., & Hardt, J. (2006). Symptoms, course, and complications of abdominal attacks in hereditary angioedema due to C1 inhibitor deficiency. *American Journal of Gastroenterology*, 101(3), 619-627. <https://www.researchgate.net/deref/http%3A%2F%2Fdx.doi.org%2F10.1111%2Fj.1572-0241.2006.00492.x>
- Bork, K., Meng, G., Staubach, P., & Hardt, J. (2006). Hereditary Angioedema: new findings concerning symptoms, affected organs, and course. *American Journal of Medicine*, 119(3), 267-274. <https://doi.org/10.1016/j.amjmed.2005.09.064>
- Bork, K., Frank, J., Gründt, B., Schlattmann, P., Nussberger, J., & Kreuz, W. (2007). Treatment of acute edema attacks in hereditary angioedema with a bradykinin receptor-2 antagonist (Icatibant). *Journal of Allergy and Clinical Immunology*, 119(6), 1497-1503. <https://doi.org/10.1016/j.jaci.2007.02.012>
- Bork, K., Bygum, A., & Hardt, J. (2008). Benefits and risks of danazol in hereditary angioedema: a long-term survey of 118 patients. *Annals Allergy Asthma Immunology*, 100(2), 153-161. [https://doi.org/10.1016/s1081-1206\(10\)60424-3](https://doi.org/10.1016/s1081-1206(10)60424-3)
- Bork, K., Wulff, K., Witzke, G., & Hardt, J. (2017). Treatment for hereditary angioedema with normal C1-INH and specific mutations in the F12 gene (HAE-FXII). *Allergy*, 72, 320-324. <https://doi.org/10.1111/all.13076>
- Bouillet, L., Longhurst, H., Boccon-Gibod, I., Bork, K., Bucher, C. Bygum, A. ... Cicardi, M. (2008). Disease expression in women with hereditary angioedema. *American Journal of Obstetrics and Gynecology*. 199(5), 484.e1-e4. <https://doi.org/10.1016/j.ajog.2008.04.034>

- Bowen, T., Cicardi, M., Bork, K., Zuraw, B., Frank, M., Ritchie, B., ... Zhi, Y. X. (2008). Hereditary angioedema: a current state-of-the-art review, VII: Canadian Hungarian 2007 International Consensus Algorithm for the Diagnosis, Therapy and Management of Hereditary Angioedema. *Annals of Allergy, Asthma & Immunology* 100, S30-S40.
[https://doi.org/10.1016/s1081-1206\(10\)60584-4](https://doi.org/10.1016/s1081-1206(10)60584-4)
- Boyle, R.J., Nikpour, M., & Tang, M. L. (2005). Hereditary angio-oedema in children: a management guideline. *Pediatric Allergy and Immunology*, 16(4), 288-294. <https://doi.org/10.1111/j.1399-3038.2005.00275.x>
- Brickman, C. M., Tsokos, G. C., Balow, J. E., Lawley, T. J., Santaella, M., Hammer, C.H., & Frank, M.M. (1986). Immunoregulatory diseases associated with hereditary angioedema: 1. Clinical manifestations of autoimmune disease. *Journal of Allergy and Clinical Immunology*, 77(5), 749–757. [https://doi.org/10.1016/0091-6749\(86\)90424-0](https://doi.org/10.1016/0091-6749(86)90424-0)
- Busse, P. J., Christiansen, S. C., Birmingham, J. M., Overbey, J. R., Banerji, A., Otani, I. M., ... Zuraw, B. L. (2019). Development of a health-related quality of life instrument for patients with hereditary angioedema living in the United States. *Journal of Allergy and Clinical Immunology in Practice*. 7, 1679-1683. <https://doi.org/10.1016/j.jaip.2018.11.042>
- Bygum, A., Andersen, K. E., & Mikkelsen, C. S. (2009). Self-administration of intravenous C1-inhibitor therapy for hereditary angioedema and associated quality of life benefits. *European Journal of Dermatology*, 19(2), 147-151. <https://www.jle.com/10.1684/ejd.2008.0603>
- Campbell, D. J., Krum, H., & Esler, M. D. (2005). Losartan increases bradykinin levels in hypertensive humans. *Circulation*, 111(3), 315-320.
<https://doi.org/10.1161/01.CIR.0000153269.07762.3B>
- Chinniah, N., & Katelaris, C.H. (2009). Hereditary angioedema and pregnancy. *Australian and New Zealand Journal of Obstetrics and Gynaecology*, 49(1), 2-5. <https://doi.org/10.1111/j.1479-828X.2008.00945.x>
- Cicardi, M., Bergamaschini, L., Cugno, M., Hack, E., Agostoni, G. & Agostoni, A. (1991). Long-term treatment of hereditary angioedema with attenuated androgens: a survey of a 13-year experience. *Journal of Allergy and Clinical Immunology*, 87(4), 768-773.
[https://doi.org/10.1016/0091-6749\(91\)90120-D](https://doi.org/10.1016/0091-6749(91)90120-D)
- Cicardi, M., Castelli, R., Zingale, L.C., & Agostini, A. (1997). Side effects of long-term prophylaxis with attenuated androgens in hereditary angioedema: comparison of treated and untreated patients. *Journal of Allergy and Clinical Immunology*, 99(2), 194-196.
[https://doi.org/10.1016/S0091-6749\(97\)70095-2](https://doi.org/10.1016/S0091-6749(97)70095-2)

- Cicardi, M., Zingale, L., Pappalardo, E., Folcioni, A., & Agostoni, A. (2003). Autoantibodies and lymphoproliferative diseases in acquired C1-inhibitor deficiencies. *Medicine (Baltimore)* 82(4), 274–281. <https://doi.org/10.1097/01.md.0000085055.63483.09>
- Craig, T., Zuraw, B., Longhurst, H., Cicardi, M., Bork, K., Grattan, C., ... Jacobs, I. (2019). Long-Term Outcomes with Subcutaneous C1-Inhibitor Replacement Therapy for Prevention of Hereditary Angioedema Attacks. *Journal of Allergy and Clinical Immunology in Practice*, 7(6), 1793–1802. <https://doi.org/10.1016/j.jaip.2019.01.054>
- Davis, A. E. (2008). Hereditary angioedema: a current state-of-the-art review, III: mechanisms of hereditary angioedema. *Annals of Allergy Asthma and Immunology*, 100 (1 Suppl 2), S7-12. [https://doi.org/10.1016/S1081-1206\(10\)60580-7](https://doi.org/10.1016/S1081-1206(10)60580-7)
- Deald, G., & Bork, K. (2006). Missense mutations in the coagulation factor XII (Hageman factor) gene in hereditary angioedema with normal C1 inhibitor. *Biochemical and Biophysical Research Communications*, 343(4), 1286-1289. <https://doi.org/10.1016/j.bbrc.2006.03.092>
- Donaldson, V. H., & Evans, R. R. (1963). A biochemical abnormality in hereditary angioneurotic edema: absence of serum inhibitor of C' 1-esterase. *American Journal of Medicine*, 35, 37-44. [https://doi.org/10.1016/0002-9343\(63\)90162-1](https://doi.org/10.1016/0002-9343(63)90162-1)
- Donaldson, V. H., Rosen, F. S. (1964). Action of complement in hereditary angioedema: the role of C'1-esterase. *Journal of Clinical Investigation*, 4, 2204–2213. <https://doi.org/10.1172/JCI105094>
- Eidelman, F. J. Eidelman (2010). Hereditary angioedema: New therapeutic options for a potentially deadly disorder. *BMC Hematology* volume 10: 3. <https://doi.org/10.1186/1471-2326-10-3>
- Farkas, H., Harmat, G., Füst, G., Varga, L., & Visy, B. (2002). Clinical management of hereditary angio-oedema in children. *Pediatric Allergy and Immunology*. 13(3), 153-161. <https://doi.org/10.1034/j.1399-3038.2002.01014.x>
- Farkas, H., Varga, L., Szeplaki, G., Visy, B., Harmat, G., & Bowen, T. (2007). Management of hereditary angioedema in pediatric patients. *Pediatrics*, 120(3), e713-722. <https://doi.org/10.1542/peds.2006-3303>
- Farkas, H., Czaller, I., Csuka, D., Vas, A., Valentin, S., Varga, ... & Karádi I. (2010). The effect of long-term danazol prophylaxis on liver function in hereditary angioedema-a longitudinal study. *Eur J Clin Pharmacol*, 66(4), 419-42. <https://link.springer.com/article/10.1007/s00228-009-0771-z>

- Frank, M. M., Gelfand, J.A., & Atkinson, J.P. (1976). Hereditary angioedema: the clinical syndrome and its management. *Annals of Internal Medicine*, 84, 580-93. <https://doi.org/10.7326/0003-4819-84-5-580>
- Frank, M. M. (1979). Effect of sex hormones on the complement-related clinical disorder of hereditary angioedema. *Arthritis and Rheumatism*, 22(11), 1295–1299. <https://doi.org/10.1002/art.1780221118>
- Frank, M. M. (2000). Urticaria and angioedema in Goldman, L. & Bennett, J. C., (ed) *Cecil Textbook of Medicine*. 21 ed. W. B. Saunders Co 1440-1445.
- Frank, M. M. (2006). Hereditary angioedema: the clinical syndrome and its management in the United States. *Immunology and Allergy Clinics of North America*, 26(4), 653-668. <https://doi.org/10.1016/j.iac.2006.09.005>
- Frank, M. M. (2008). Hereditary angioedema: a current state-of-the-art review, VI: novel therapies for hereditary angioedema. *Annals of Allergy Asthma and Immunology*, 100(1 Suppl 2), S23-29. [https://doi.org/10.1016/S1081-1206\(10\)60583-2](https://doi.org/10.1016/S1081-1206(10)60583-2)
- Gompels, M. M., Lock, R. J., Abinun, M., Bethune, C. A., Davies, G., Grattan, C., ... Watters, D. (2005). C1 inhibitor deficiency: consensus document. *Clinical and Experimental Immunology*, 139(3), 379-394. <https://doi.org/10.1111/j.1365-2249.2005.02726.x>
- Graves, R. (1843). Clinical lectures on the practice of medicine, in Major, R.H., ed. (1955). *Classic Descriptions of Disease*. 3rd Ed. Charles C. Thomas Pub Ltd, 623- 624.
- Jurado-Palomo, J., Muñoz-Caro, J. M., López-Serrano, M. C., Prior, N., Cabañas, R., Pedrosa, M., Burgueño, M., & Caballero, T. (2013). Management of Dental-Oral Procedures in Patients with Hereditary Angioedema due to C1 Inhibitor Deficiency. *The Journal of Investigational Allergology and Clinical Immunology*, 23(1), 1-6. [http://www.jaci.org/issues/vol23issue1/1.pdf](http://www.jiaci.org/issues/vol23issue1/1.pdf)
- Landerman, N. S., Webster, M. E., Becker, E. L., & Ratcliffe, H. E. (1962). Hereditary angioneurotic edema. *Journal of Allergy*, 33, 330–341. [https://doi.org/10.1016/0021-8707\(62\)90031-X](https://doi.org/10.1016/0021-8707(62)90031-X)
- Longhurst, H. J. (2005). Emergency treatment of acute attacks in hereditary angioedema due to C1 inhibitor deficiency: what is the evidence? *International Journal of Clinical Practice*, 59(5), 594-599. <https://doi.org/10.1111/j.1742-1241.2005.00352.x>
- Longhurst, H., Cicardi, M., Craig, T., Bork, K., Grattan, C., Baker, J., ... Zuraw, B. L. for COMPACT Investigators. (2017). Prevention of Hereditary Angioedema Attacks with a Subcutaneous C1 Inhibitor. *New England Journal of Medicine*, 376, 1131–40. <https://www.nejm.org/doi/pdf/10.1056/NEJMoa1613627>

- Magerl, M., Germanis, A. E., Maas, C., & Maurer, M. (2017). Hereditary Angioedema with Normal C1 Inhibitor: Update on Evaluation and Treatment. *Immunology and Allergy Clinics of North America*, 37(3), 571-584. <https://doi.org/10.1016/j.iac.2017.04.004>
- Maurer, M., Longhurst, H. J., Fabien, V., Li, H. H., & Lumry, W. R., for the IOS Study Group. (2014). Treatment of hereditary angioedema with icatibant: efficacy in clinical trials versus effectiveness in the real-world setting. *Allergy Asthma Proc*, 35(5), 377-81. <https://doi.org/10.2500/aap.2014.35.3780>
- Maurer, M., Magerl, M., Ansotegui, I., Aygoren-Pursun, E., Betschel, S., Bork, K. ... Craig, T. (2018). The international WAO/EAACI guideline for the management of hereditary angioedema – the 2017 revision and update. *World Allergy Organization Journal*, 11, Article number 5. <https://doi.org/10.1186/s40413-017-0180-1>
- Nzeako, U. C., Frigas, E., & Tremaine, W. J. (2001). Hereditary angioedema: a broad review for clinicians. *Archives of Internal Medicine*, 161, 2417-2429. <https://jamanetwork.com/journals/jamainternalmedicine/fullarticle/649449>
- O'Bier, A., Muniz, A.E., & Foster, R.L. (2005). Hereditary angioedema presenting as epiglottitis. *Pediatric Emergency Care*, 21(1), 27-30. <https://doi.org/10.1097/01.pec.0000150985.81109.0d>
- Osler, W. (1888). Hereditary angio-neurotic oedema. *American Journal of Medical Science*, 95, 362–367. [https://www.amjmedsci.org/article/S0002-9629\(15\)31692-X/fulltext](https://www.amjmedsci.org/article/S0002-9629(15)31692-X/fulltext)
- Postnikoff, I. M. & Pritzker, K. P. (1979). Hereditary angioneurotic edema: an unusual case of maternal mortality. *Journal of Forensic Sciences*, 24, 473-478. <https://doi.org/10.1520/JFS10855J>
- Prematta, M. J., Prematta, T., & Craig, T. J. (2008). Treatment of HAE with plasma-derived C1inhibitor. *Therapeutics & Clinical Risk Management*, 4(5), 1-8. <https://doi.org/10.2147/TCRM.S3172>
- Prior, N., Remor, E., Gomez-Traseira, C., Lopez-Serrano, C., Cabanas, R., Contreras, J., ... Cabereloo, T. (2012). Development of a disease-specific quality of life questionnaire for adult patients with hereditary angioedema due to C1 inhibitor deficiency (HAE- QoL): Spanish multi-centre research project. *Health Qual Life Outcomes*, 10, 82. <https://doi.org/10.1186/1477-7525-10-82>
- Quincke, H. (1882). Concerning the acute localized oedema of the skin. *Monatsh Prakt Dermatology*, 1, 129–31.

- Roche, O., Blanch, A., Caballero, T., Sastre, N., Callejo, D., & Lopez-Trascasa, M. (2005). Hereditary angioedema due to C1 inhibitor deficiency: patient registry and approach to the prevalence in Spain. *Annals of Allergy, Asthma, & Immunology*, 94, 498-503.
[https://doi.org/10.1016/S1081-1206\(10\)61121-0](https://doi.org/10.1016/S1081-1206(10)61121-0)
- Rosen, F. S., Pensky, J., Donaldson, V., & Charache, P. (1965). Hereditary angioneurotic edema: two genetic variants. *Science*, 148, 957–958. <https://doi.org/10.1126/science.148.3672.957>
- Szeplaki, G., Varga, L., Kleiber, M., Karadi, I., Romics, L., Fust, G., & Farkas, H. (2005). Adverse effects of danazol prophylaxis on the lipid profiles of patients with hereditary angioedema. *Journal of Allergy and Clinical Immunology*, 115, 864-869.
<https://doi.org/10.1016/j.jaci.2004.12.1130>
- Weller, K., Groffik, A., Magerl, M., Tohme, N., Martus, P., Krause, K., ... Maurer, M. (2012). Development and construct validation of the angioedema quality of life questionnaire. *Allergy*, 67(10), 1289–98. <https://doi.org/10.1111/all.12007>
- Weller, K., Magerl, M., Peveling-Oberhag, A., Martus, P., Staubach, P., & Maurer, M. (2016). The Angioedema Quality of Life Questionnaire (AE-QoL) - assessment of sensitivity to change and minimal clinically important difference. *Allergy*, 71, 1203–9.
<https://doi.org/10.1111/all.12900>
- Winnewisser, J., Rossi, M., Späth, P., & Bürgi, H. (1997). Type I hereditary angio-oedema. Variability of clinical presentation and course within two large kindreds. *Journal of Internal Medicine*, 241(1). <https://doi.org/10.1046/j.1365-2796.1997.76893000.x>
- Wu, M. A. Lanadelumab for the treatment of hereditary angioedema. (2019). *Expert Opinion on Biological Therapy*. 19, 1233–45. <https://doi.org/10.1080/14712598.2019.1685490>
- Zingale, L. C., Beltrami, L., Zanichelli, A., Maggioni, L., Pappalardo, E., Cicardi, B., & Cicardi, M. (2006). Angioedema without urticaria: a large clinical survey. *Canadian Medical Association Journal*, 175, 1065-1070. <https://doi.org/10.1503/cmaj.060535>
- Zuraw, B. L. & Herschbach, J., (2000). Detection of C1 inhibitor mutations in patients with hereditary angioedema. *Journal of Allergy Clinical Immunology*, 105(3), 541-546.
<https://doi.org/10.1067/mai.2000.104780>
- Zuraw, B. L. (2008). Clinical practice. Hereditary angioedema. *New England Journal of Medicine*, 359, 1027-1036. <http://doi.org/10.1056/NEJMcp0803977>
- Zuraw, B. L. (2018). Hereditary angioedema with normal C1 inhibitor: Four types and counting. *J Allergy Clin Immunol*. 141(3), 884-5. <https://doi.org/10.1016/j.jaci.2018.01.015>

Zuraw B. L., Lumry, W., Banerji, A., Aygoren-Pursun, E., Bernstein, J., Johnston, D. ... Sheridan, W. (2019). P150 oral prophylaxis with bcx7353 reduces HAE attack rates and is well-tolerated: apex-2 study results. *Annals Allergy Asthma & Immunol*, 123(5), S27.

[https://www.annallergy.org/article/S1081-1206\(19\)30843-9/fulltext](https://www.annallergy.org/article/S1081-1206(19)30843-9/fulltext)

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