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## GENETIC TESTING FOR ANGIOEDEMA REFERENCES

HEREDITARY ANGIOEDEMA (HAE) GENES				
Gene	OMIM	Exon(s)	Other Information	
C1INH Gene			Region	
SERPING1 <sup>1, 10</sup>	<a href="#">106100</a>	2-8	Coding ± 10 base pairs	
nC1INH Genes <sup>2</sup>			Screen for known HAE variant(s)	
F12 <sup>3, 11, 12</sup>	<a href="#">610618</a>	9	c.983C>A	p.Thr328Lys
			c.983C>G	p.Thr328Arg
			c.971_1018+24del72	
			c.892_909dup	
KNG1 <sup>6</sup>	<a href="#">619363</a>	10	c.1136T>A	p.Met379Lys
ANGPT1 <sup>5</sup>	<a href="#">619361</a>	2	c.355G>T	p.Ala119Ser
PLG <sup>4, 7</sup>	<a href="#">619360</a>	9	c.988A>G	p.Lys330Glu
		18	c.2183T>A	p.Val728Glu
MYOF <sup>8</sup>	<a href="#">619366</a>	7	c.651G>T	p.Arg217Ser
HS3ST6 <sup>9</sup>	<a href="#">619367</a>	2	c.430A>T	p.Thr144Ser

### REFERENCES

1. Patel G, Pongracic JA. Hereditary and acquired angioedema. Allergy Asthma Proc. 2019 Nov 1;40(6):441-445. doi: 10.2500/aap.2019.40.4267. PMID: 31690390.
2. Bork, K., Machnig, T., Wulff, K., Witzke, G., Prusty, S., Hardt, J. Clinical features of genetically characterized types of hereditary angioedema with normal C1 inhibitor: a systematic review of qualitative evidence. Orphanet J. Rare Dis. 15: 289, 2020. [PubMed: [33059692](#), [images](#), [related citations](#)]
3. Cichon, S., Martin, L., Hennies, H. C., Muller, F., Van Driessche, K., Karpushova, A., Stevens, W., Colombo, R., Renne, T., Drouet, C., Bork, K., Nothen, M. M. Increased activity of coagulation

- factor XII (Hageman factor) causes hereditary angioedema type III. *Am. J. Hum. Genet.* 79: 1098-1104, 2006. [PubMed: [17186468](#), [images](#), [related citations](#)]
4. Farkas, H., Doczy, A., Szabo, E., Varga, L., Csuka, D. Screening for plasminogen mutations in hereditary angioedema patients. *Genes* 12: 402, 2021. [PubMed: [33799813](#), [related citations](#)]
  5. Zuraw, B. L. Hereditary angioedema with normal C1 inhibitor: four types and counting. *J. Allergy Clin. Immun.* 141: 884-885, 2018. [PubMed: [29410040](#), [related citations](#)]
  6. Bork, K., Wulff, K., Rossmann, H., Steinmuller-Magin, L., Braenne, I., Witzke, G., Hardt, J. Hereditary angioedema cosegregating with a novel kininogen 1 gene mutation changing the N-terminal cleavage site of bradykinin. (Letter) *Allergy* 74: 2479-2544, 2019. [PubMed: [31087670](#), [related citations](#)]
  7. Loules, G., Parsopoulou, F., Zamanakou, M., Csuka, D., Bova, M., Gonzalez-Quevedo, T., Psarros, F., Porebski, G., Speletas, M., Firinu, D., del Giacco, S., Suffritti, C., Makris, M., Vatsiou, S., Zanichelli, A., Farkas, H., Germanis, A. E. Deciphering the genetics of primary angioedema with normal levels of C1 inhibitor. *J. Clin. Med.* 9: 3402, 2020. [PubMed: [33114181](#), [related citations](#)]
  8. Ariano, A., D'Apolito, M., Bova, M., Bellanti, F., Loffredo, S., D'Andrea, G., Intrieri, M., Petraroli, A., Maffione, A. B., Spadaro, G., Santacroce, R., Margaglione, M. A myoferlin gain-of-function variant associates with a new type of hereditary angioedema. (Letter) *Allergy* 75: 2989-2992, 2020. [PubMed: [32542751](#), [related citations](#)]
  9. Bork, K., Wulff, K., Mohl, B. S., Steinmuller-Magin, L., Witzke, G., Hardt, J., Meinke, P. Novel hereditary angioedema linked with a heparan sulfate 3-O-sulfotransferase 6 gene mutation. *J. Allergy Clin. Immun.* 148: 1041-1048, 2021. [PubMed: [33508266](#), [related citations](#)]
  10. Wilkerson RG, Moellman JJ. Hereditary Angioedema. *Emerg Med Clin North Am.* 2022 Feb;40(1):99-118. doi: 10.1016/j.emc.2021.09.002. Epub 2021 Oct 29. PMID: 34782094.
  11. Bork K, Wulff K, Witzke G, Hardt J. Hereditary angioedema with normal C1-INH with versus without specific F12 gene mutations. *Allergy.* 2015 Aug;70(8):1004-12. doi: 10.1111/all.12648. Epub 2015 May 22. PMID: 25952149.
  12. Kiss N, Barabás E, Várnai K, Halász A, Varga LÁ, Prohászka Z, Farkas H, Szilágyi Á. Novel duplication in the F12 gene in a patient with recurrent angioedema. *Clin Immunol.* 2013 Oct;149(1):142-5. doi: 10.1016/j.clim.2013.08.001. Epub 2013 Aug 9. PMID: 23994767.