Genetics of Hidradenitis Suppurativa

Airon Li* and Rudolph E. Tanzi*

Genetics and Aging Research Unit, Department of Neurology, Massachusetts General Hospital and Harvard Medical School, Charlestown, MA, US

*Corresponding authors: Airon Li MD PhD, Genetics and Aging Research Unit Massachusetts General Hospital, Harvard Medical School, 114, 16th Street MA 02129, US; Tel: 6177249397; E-mail: ali3@mgh.harvard.edu

Rudolph E. Tanzi PhD, Genetics and Aging Research Unit Massachusetts General Hospital, Harvard Medical School, 114, 16th Street MA 02129, US; Tel: 6177266845; Fax: 617-724-1823; E-mail: rtanzi@mgh.harvard.edu

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Abstract

Hidradenitis Suppurativa (HS), also named acne inversa, which is a common chronic inflammatory skin disorder characterized clinically by painful lumps, abscesses and scarring. Thirty-five unique mutations in patients with HS have been identified in three of the genes that encode members of the γ-secretase complex: nicastrin (NCSTN), presenilin 1 (PSEN1), and presenilin enhancer 2 (PSENEN) as well as in POGLUT1, an Endoplasmic Reticulum (ER) O-glucosyltransferase that is involved in Notch signaling. This review summarizes research updates on genetics of HS.

Keywords: Hidradenitis suppurativa, γ-secretase, nicastrin, presenilin

Introduction of HS

Hidradenitis Suppurativa (HS), also named acne inversa, is a common chronic inflammatory skin disorder characterized clinically by painful lumps, abscesses and scarring (OMIM # 142690). The prevalence of HS in the population is 0.10%, or 98 per 100,000 persons in the United States (US) [1,2] and three times more common in female patients (73.8% women) than male patients (26.2% men), 3-fold greater in African Americans and 2-fold greater in biracial populations than in the overall population [1]. Antibiotics, anti-inflammatory regiments, acne washes and medicines, and surgical procedure are the premirary current treatment options [3]. Major surgery demonstrated improvements in the HS patients’ overall work and daily activity impairment [4]. However, the disease progression often causes scars leading immobility, markedly affecting quality of life in severe patients who have poor responses to treatments [5].

The etiology of HS is associated with multi-factorals including genetics and others. HS increased an independent risk of all-cause mortality [6]. Obesity, smoking, family history and environmental factors such as diet, are known to be associated with the HS disease pathogenesis. Obesity is linked to skin barrier function, sebaceous glands and sebum production, sweat gland, lymphatics, and collagen structure and function, wound healing, microcirculation and macrocirculation [7]. Obesity and smoking increase the HS incidence [8] [9]. HS patients classified as Hurley III HS were 28% more likely to be smokers and obese [10] and four times more likely to be obese compared to the general population by meta-analysis of case-control studies in Asia, Europe, and the US [11]. One-third (31%) of the HS patients who eliminated smoking or made dietary alterations including a reduction in gluten, dairy, refined sugars, tomatoes, or alcohol showed improvement in HS clinical symptoms [12]. Patients with HS were at higher risk for long-term opioid use compared with controls [13].

HS lesion counts are increased with low serum zinc and vitamin D levels. Supplementation of zinc, vitamin D, vitamin B12, or exclusion of dairy or brewey’s yeast reduced lesion resolution. Bariatric surgery often causes weight loss which may lead to HS improvement but often results in more severe malnutrition that worsens or even leads to new HS onset post bariatric surgery [11]. The complement (C) system was found to be significantly down-regulated in the HS skin and blood transcriptomes and the HS blood proteome [4]. Porphyromonas species, which are able cleave inactive C5 into C5a, have been identified in the HS microbiome. C5a levels in serum and tissue correlate with disease activity and degree of neutrophilic infiltrates in HS, suggesting that complement inhibition is a promising and potential therapeutic target for HS [82]. HS lesions showed 83% bacterial culture anaerobes comprised to 53% of control samples, and milleri group streptococci and actinomycetes in 33% and 26% of cases, respectively [83]. Microarray analysis demonstrated that HS lesional skin samples had significantly decreased expression of enzymes involved in generating ceramide and sphingomyelin, increased expression of enzymes that catabolize ceramide to sphingosine, and increased expression of enzymes involved in converting ceramide to galactosylceramide and gangliosides, which suggests that sphingolipid metabolism is altered in HS lesional skin compared with normal skin [86]. In HS patients, the serum and HS skin lesion levels of chitinase-3-like protein 1 (YKL-40) were significantly elevated, suggesting that YKL-40 may be one of the biomarkers of HS [87].

HS patients demonstrate a significantly higher heart rate in the HS groups than in the population [14]. HS often co-existed with psoriasis. Compared to patients with psoriasis alone, HS patients with psoriasis were significantly younger and had a higher prevalence of obesity and smoking [15].

Macrophages in HS infiltrates release a variety of pro-inflammatory cytokines such as interleukins and tumor necrosis factor α (TNFα),...
exacerbating the inflammation. Obesity and smoking contribute to macrophage dysfunction [9]. Elevated expression of TNFα has been identified in skin lesions, such as skin tunnels, of HS patients along with a clustering of interleukins (IL-8, IL-16, IL-1α and IL-1β) [68] [69]. Gene-sets related to Notch signalling and Interferon pathways were differentially activated in HS lesional compared to non-lesional skin [80].

Adalimumab is a TNFα inhibitor which has been used in both USA and Europe for treating HS patients. Adalimumab reduced flare, showed a higher efficacy on nodules-abscesses than on draining tunnels and increased the number of patients achieving a Hidradenitis Suppurativa Clinical Response [91]. By a Genome-Wide Association Study (GWAS) analysis one single Linkage Disequilibrium (LD) block in the BCL2 gene was significantly associated with adalimumab response (lead Single-Nucleotide Polymorphism [SNP] rs 59532114). Meanwhile, a correlation of the most strongly associated SNP minor allele with increased BCL2 gene and protein expression in hair follicle tissues was observed with bioinformatic analysis and functional genomics experiments [66]. HLA alleles may affect the treatment response in HS patients treated with adalimumab. There were three protective HLA alleles (HLA-DQB1*05, HLA-DRB1*01, and HLA-DRB1*07) less prevalent and two risk HLA alleles (HLA-DRB1*03 and HLA-DRB1*011) more abundant in HS patients developing anti-drug antibodies to adalimumab than these not [67].

**Genes Linked to HS**

Genetics is associated with the pathogenesis of HS. One third of HS patients have a family history with an autosomal dominant inheritance trait [16] which pattern suggests a single gene disorder. Thirty-five unique mutations in patients with familial or sporadic HS have been found in genes encoding three of the four genes comprising the γ-secretase complex: nicastrin (NCSTN), presenilin 1 (PSEN1), presenilin enhancer 2 (PSENEN) [17] [18] [19] [20] [21] [22] [23]

### Table 1: Mutation spectrum of NCSTN, PSEN1, PSENEN and POGLUT1 in HS patients

<table>
<thead>
<tr>
<th>ID</th>
<th>Mutation category</th>
<th>Nucleotide Change</th>
<th>Amino Acid Change</th>
<th>TM</th>
<th>Ethnic origin</th>
<th>Reference</th>
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<td>1</td>
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<td>p.V75I</td>
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<tr>
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<td>p.P211R</td>
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<tr>
<td>4</td>
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<td>p.Q216P</td>
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<td>Chinese</td>
<td>18</td>
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<tr>
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<td>p.A315V</td>
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<td>9</td>
<td>Nonsense</td>
<td>c. 497C&gt;A</td>
<td>p.S166X</td>
<td>No</td>
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<td>18</td>
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<tr>
<td>10</td>
<td>Nonsense</td>
<td>c. 1258C&gt;T</td>
<td>p.Q420X</td>
<td>No</td>
<td>Chinese</td>
<td>20</td>
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<tr>
<td>11</td>
<td>Nonsense</td>
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<td>p.R434X</td>
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<td>Nonsense</td>
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<td>p.Y565X</td>
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<td>p.Q568X</td>
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<td>Japanese</td>
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<td>c. 1799delTG</td>
<td>p.L600X</td>
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<tr>
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<td>Frameshift</td>
<td>c. 210_211delAG</td>
<td>p.T706X18</td>
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<tr>
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<td>p.Q163S63X39</td>
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<tr>
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<td>Frameshift</td>
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<td>p.C230P65X41</td>
<td>No</td>
<td>Indian</td>
<td>17</td>
</tr>
<tr>
<td>18</td>
<td>Frameshift</td>
<td>c. 1752delG</td>
<td>p.E584Q65X44</td>
<td>No</td>
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<tr>
<td>19</td>
<td>Frameshift</td>
<td>c. 1768A&gt;G</td>
<td>p.S590A63X3</td>
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<td>Caucasian</td>
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<tr>
<td>20</td>
<td>Frameshift</td>
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<td>p.S638S6X1</td>
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<tr>
<td>21</td>
<td>Splice Site</td>
<td>c. 582+1delG</td>
<td>p. F1456X54</td>
<td>No</td>
<td>Japanese</td>
<td>26</td>
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<tr>
<td>22</td>
<td>Splice Site</td>
<td>c. 996+7+G&gt;A</td>
<td>p.L282_G332del</td>
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<td>17</td>
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<tr>
<td>23</td>
<td>Splice Site</td>
<td>c. 1101+1+G&gt;A</td>
<td>p.E333_Q367del</td>
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<tr>
<td>24</td>
<td>Splice Site</td>
<td>c. 1101+1+T&gt;A+G</td>
<td>p.E333_Q367del</td>
<td>Yes</td>
<td>African</td>
<td>17</td>
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<tr>
<td>25</td>
<td>Splice Site</td>
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<td>p.Q393S_X9</td>
<td>No</td>
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<td>26</td>
<td>Splice Site</td>
<td>c. 1551+1+G&gt;A</td>
<td>p.A486_T517del</td>
<td>No</td>
<td>Chinese</td>
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<tr>
<td>27</td>
<td>Frameshift</td>
<td>c. 725delC</td>
<td>p. P424L6X11</td>
<td>Chinese</td>
<td>25</td>
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<tr>
<td>28</td>
<td>Frameshift</td>
<td>c. 656delG</td>
<td>p. F231L6X46</td>
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<td>Frameshift</td>
<td>c. 66_67insG</td>
<td>p. F231L6X98</td>
<td>Caucasian</td>
<td>17</td>
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</tr>
<tr>
<td>30</td>
<td>Frameshift</td>
<td>c. 279delC</td>
<td>p. P946S5X1</td>
<td>Chinese</td>
<td>24</td>
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<td>p. Y56_101Fdel</td>
<td>Caucasian</td>
<td>18</td>
<td></td>
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<tr>
<td>33</td>
<td>Missense</td>
<td>c. 1947T&gt;G</td>
<td>p. L65R</td>
<td>Chinese</td>
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<tr>
<td>34</td>
<td>Nonsense</td>
<td>c. 814C&gt;T</td>
<td>p. R272</td>
<td>Caucasian</td>
<td>26</td>
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<tr>
<td>35</td>
<td>Splicing</td>
<td>c. 430_1G&gt;A</td>
<td>p. K246_392Ldel</td>
<td>Caucasian</td>
<td>25</td>
<td></td>
</tr>
</tbody>
</table>

**NCSTN**

**PSEN1**

**PSENEN**

**POGLUT1**

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and c.1352+1 G>A (experimental confirmed) [27] lose the TM NCSTN of other nonsense, frameshift mutations and c.582+1delG and 4 splicing site mutations) retain the TM region, while 61% (16/26) of NCSTN mutations affect important substrate recruitment structures. 50% (3/6) of the NCSTN splicing site mutations affect substrate recruitment [34].

**Post Translation of HS-Linked Genetic Mutations**

NCSTN mutation NS565X occurs on a tyrosine phosphorylation site and R434X occurs on a glycosylation site. NCSTN-R434X disrupts the protein immediately before Asn55 and Asn435 [40]. 21% (5 of 24) of the NCSTN mutations, NCSTN-P211R, L600X, C230PfsX31, P590AfsX3 and F145fs_X54 occur at cysteine residues participating in disulfide bonds [41] [42]. Six potential NCSTN ubiquitination sites are predicted: K78, T127, K386, K403, K591 and K597. Six residues in NCSTN undergo sumoylation: G146, S341, K386, P423, T459, and D476. NCSTN-P590AfsX3 occurs immediately before the predicted ubiquitination site K591 and abolishes two ubiquitination sites - K591 and K597. F145fs_X54 abolishes sumoylation site G146. Both NCSTN - E333_Q367del and E333_Q367del abolish sumoylation site S341. NCSTN-T706fsX18 and R117X abolish all the ubiquitination and sumoylation sites and C159X and S166X abolish four of the six ubiquitination sites and five of the six sumoylation sites [34]. The C-terminal end frameshift mutation NCSTN-E854DfsX44 resulted in a striking 3D structural change suggesting that this mutation is likely located at a critical site for NCSTN conformation [34]. Ubiquitination and sumoylation are involved in post-translational modification. A large number of NCSTN mutations affect predicted ubiquitination and sumoylation sites, suggesting that post-translational modification might contribute to HS pathogenesis.

**HS-Linked Mutational Effect**

HS associated mutations in NCSTN are predicted to cause a loss of function as a result of frameshift and premature translation termination and a loss of the TM domain, to affect NCSTN substrate recruitment sites, to cause a loss or creation of new ligand binding sites, and to alter post-translational modifications and disulfide bonds [41] [42], all of which support the notion that the NCSTN mutations result in significantly reduced levels of NCT and reduced γ-secretase-mediated processing of Notch and signaling in the skin [43]. Silencing of the keratinocyte NCSTN by CRISPR-Cas9 in both the keratinocyte cell line HEK001 and an embryonic kidney cell line HEK293 showed a significantly increased expression of genes related to the type I interferon response pathway [44]. NCSTN Wild Type (WT) were upregulated in myeloid cells including monocytes, macrophages and non-lymphoid dendritic cells [35]. NCSTN knockdown in HaCaT cells impaired γ-secretase activity and proliferation and differentiation of keratinocytes. Expression levels of several γ-secretase substrates involved in the Notch pathway were significantly attenuated in...
NCSTN-silencing HaCaT cells and the lesion from a HS patient. Phosphoinositide 3-kinase (PI3K) as well as AKT and its activated form pAKT were markedly elevated in NCSTN-silencing HaCaT cells [23]. NCSTN mutations led to decreased miR-30a-3p levels, which negatively regulated RAB31 expression. Moreover, enhanced RAB31 levels accelerated degradation of activated EGFR, leading to abnormal differentiation in keratinocytes. Familial HS patients and mouse knocked out for Ncstn showed impaired EGFRsignaling and epidermal differentiation [45].

However, testing four NCSTN-missense mutations, V75I, D185N, P211R, and Q216P for their effects on mediating Notch processing and signaling demonstrated a vague role of HS-linked NCSTN mutations in HS pathogenesis. The NCSTN-V75I, D185N, and P211R mutants can function in Notch signaling in vivo; in contrast mutant Q216P failed to rescue Notch processing and nuclear signaling [46]. Mouse models where components of the γ-secretase with resultant Notch dysregulation have been knocked out have resulted in the development of dermal cysts and histological features of follicular occlusion [21] [47] although these models rapidly developed multiple squamous cell carcinomas, which is not consistent with the typical progression of HS [47]. These findings suggest that although NCSTN-V75I, D185N, and P211R and some other NCSTN mutations have a significant role in the pathogenesis of the disease, this role is through a mechanism(s) other than impaired Notch signaling.

A single frameshift PSEN1-P242LfsX11 mutation is predicted to truncate the PS1 protein after the 5th TM domain at the cytosolic region of the N terminal, which would markedly alter the 3D structure of PS1. PSENEN contains three TMs, at amino acid positions 18-38, 60-80 and 85-101. The PSENEN N-termius is cytoplasmic, followed by two short helices that dip into the membrane [40]. All the PSENEN mutations occur within TM regions: frameshifting mutations F23LfsX46 and F23VfsX98 delete all 3 TM regions, while P49SfsX51 disrupts TM region 3. Nonsense Y56-101Pdel and c.167-2A>G splicing site mutations lead to similar disruptions of TM regions 2 & 3. The missense mutation PSENEN-L65R lays in the TM 2 region and is predicted to be deleterious. POGLUT1 is located in the lumen of the endoplasmic reticulum. Both POGLUT1-R272* and C.430-1G>A, K246* lead to an early termination of protein synthesis. POGLUT1-R272* is located in the C-terminal domain and results in a truncated form of POGLUT1 with partial loss of the C-terminal domain. The splicing site c.430-1G>A mutation was identified in exon 4 of the POGLUT1 gene in patients with HS and DDS syndrome, which potentially generates aberrant splicing with loss of functionality [33]. POGLUT1 is predicted to possess 17 ligand binding sites of interactions with chain A. Hydrogen bonds include A.Y117, A.S152, A.R158, A.R158, A.D196, A.V197, A.L199, A.V214, A.A215, A.A215, A.S217, A.F218, A.R219, A.R219 and salt bridges: A.R158 and A.R219. Both POGLUT1- c.430-1G>A (K246*) or R272* completely abolish ligand binding function and show significant alteration of global quality estimate by Qualitative Model Energy Analysis (QMEAN) values: POGLUT1-WT: -71; POGLUT1- c.430-1G>A (K246*): 0.90; and R272* 0.45, indicating a greater deviation in mutant forms from the POGLUT1-WT [34].

A higher and prolonged TNFa expression and differential gene expression of four cytokine or chemokines than that of PS1-WT in response to LPS stimulation was observed in overexpression of the HS-associated PSEN1 mutation PSEN1-P242LfsX11 in PMA-differentiated macrophages [34]. Of the overexpressing PSEN1-WT and PSEN1-P242LfsX11 induced under-expressed genes [34], LIF and CSF2 are essential for the proliferation and differentiation of hematopoietic progenitor cells into granulocytes and macrophages [48] [49], IL12 is critical for the activation and maintenance of immune responses [50], and BMP2 regulates stem cell activation in the process of hair follicle regeneration in the dermis [51]. The increased expression of proinflammatory TNFa and the decreased expression of LIF, IL12B, CSF2, BMP2 and other genes associated with the overexpression of PSEN1-P242LfsX11 may promote inflammatory processes, impair the activation/maintenance of immune cells and reduce hair follicle regeneration [34]. HS patients with a PSEN1 mutation may benefit greatly from TNFa inhibiting agents such as infliximab, adalimumab, rituximab, and ustekinumab, in particular after anti-inflammatory regimens fail to control the disease process.

PSEN1 has pleiotropic nature [52]. PSEN1 is linked to early-onset familial Alzheimer’s Disease (AD) (OMIM # 104300), a neurodegenerative disorder and the most common form of dementia in the elderly [53]. A single frameshif PSEN1-P242LfsX11 mutation was detected in familial HS patients [21]. More than 185 missense or inframe deletion mutations and promoter variants in PSEN1 have previously been found in patients with familial AD (http://www. alz. org/) and sporadic Dilated Cardiomyopathy (DCM) [54], and 685 genes have been associated with AD (www.alz.org). The familial HS patients with PSEN1-P242LfsX11 mutation did not show the symptoms of AD [21]. Significant differential expression of ErbB4, SCNB1, and T1e1 was observed in HS lesional skin, and of EphB2, EPHB4, KCNE1, LRP6, MUSK, SDC3, Sortilin1 were observed in blood specific to AD [55]. AD-associated PSEN1 mutations alters the γ-secretase cleavage of β-APP to increase β/40 ratio resulting in β plaque formation and related AD pathology [21]. Overexpression or silencing of presenilin caused cardiac dysfunction in Drosophila [56]. Overexpression of PSEN1-P242LfsX11 in zebrafish embryos enhanced Notch signaling but did not affect γ-secretase cleavage of APP [57], which suggests that the involvement of the PSEN1 mutation in HS pathogenesis also has a mechanism that is independent of γ-secretase activity. Different from the effectiveness of administration of TNFa inhibitor Adalimumab in the treatment of HS patients, administration of the TNFa modulator etanercept in AD patients demonstrated no apparent effect on cognitive functioning, though TNFa has been implicated in the pathogenesis of AD [58] [59]. In AD patients, only one side of each TM helix in PS1 is affected, the hot spot of Leu219, Glu222, Leu226, Ser230, Met233, and Phe237 are placed on the same side of TM5 [40] while the HS-linked PSEN1-P242LfsX11 is on the other side of TM5 in PS1. This distribution or structure of AD-linked PSEN1 mutation is significantly different from HS-linked PSEN mutations which may indicate functional importance.

POGLUT1 is an Endoplasmic Reticulum (ER) O-glycosyltransferase that adds glucose moieties to serine residues in EGF-like repeats, such as NOTCH intracellular domain [60]. Mutations in POGLUT1, including W4X, R218X, R279PfsX3 and
miRNAs may be potential disease biomarkers and therapeutic targets. Significant epigenetic modifications were observed in HS skin lesions. mRNA of all the studied genes were significantly under-expressed in lesional HS skin compared to healthy controls, suggesting that epigenetic changes occur in HS tissue and that aberrant expression of the DNA hydroxymethylation regulators may play a role in the pathogenesis of HS. HS was associated with a 1.69-fold increased odds of diabetes; however, the absolute risk difference was small and is probably not clinically relevant. A significant overexpression of miRNA-155-5p, miRNA-223-5p, miRNA-31-5p, miRNA-21-5p, and miRNA-146a-5p was observed in lesional HS skin compared to healthy controls, suggesting that these miRNAs may be potential disease biomarkers and therapeutic targets for HS.

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