

## Supplemental Data

### Nonsense Mutations in *AAGAB*

### Cause Punctate Palmoplantar Keratoderma

### Type Buschke-Fischer-Brauer

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## Material and Methods

### *Standard Procedure for Indirect Immunofluorescence Microscopy*

After briefly rinsing in PBS, cryostat sections were fixed in acetone (-20°C; 5min), blocked with 5% normal goat serum in phosphate-buffered saline (PBST; PBS + 0.001% Triton-X100). Primary antibodies were applied for 1h and after rinsing in PBS, incubation with secondary antibodies were done for 30min. After washing in PBS, sections were dried and mounted. To prevent possible losses of soluble antigens i.e. transcription factors during the immunolocalization procedures using sections of frozen tissues, different fixation and incubation protocols were used (protocols 2 and 3)<sup>1</sup>. Briefly, cryosections were very shortly rinsed in PBS, supplemented with 2mM MgCl<sub>2</sub> and following deviations were applied: Instead of PBST only PBS was used and all antibody incubation steps were shortened to 15 min and the washing steps to 3 min under shaking ("short protocol") or the initial fixation was done in freshly prepared phosphate-buffered 2% formaldehyde in PBS, 15 min, RT, followed by rinsing in PBS, blocking in 50 mM NH<sub>4</sub>Cl (freshly prepared, 10 min, RT), rinsing in PBST ("formaldehyde protocol").<sup>1</sup> Thereafter the standard procedure was followed.

Immunofluorescence was documented with a Zeiss AxioPhot microscope equipped with an AxioCam MRm camera and AxioVision imaging software (Carl Zeiss Oberkochen/ Jena, Germany).

### *Antibodies*

Specific antibodies against human *AAGAB* were: Purified mouse polyclonal antibodies (FLJ11506; Catalog: H00079710- B01P; dilution 1:20; Abnova, Taipei, Taiwan), rabbit polyclonal antibodies (FLJ11506; Catalog: NBP1-87905; dilution 1:50) and (FLJ11506; catalog: NBP1-87906; dilution 1:100; both from Novus Biologicals, Cambridge, UK). For positive control of tissue antigenicity, anti-human Keratin K1 made in guinea pigs (diluted 1:2000; DKFZ by L.L.) was used. Secondary antibodies (IgG or IgG+IgM) used for IIF were: Goat anti-rabbit-, anti-guinea pig, -anti-mouse, coupled to Cy3 (1:500, red fluorescence, Molecular Probes).

The *AAGAB* antisera were partially characterized for specificity by the providers mainly by Western blotting, but little was known for *AAGAB* immunohistochemical staining

procedures. We therefore tested all sera for appropriate dilutions and fixation procedures. As our standard fixation procedure including permeabilization of cells, was not successful we used a previously described protocol for soluble materials (see supplement).<sup>1-2</sup> Formaldehyde fixation gave the best results and was used throughout the study.

## References

1. Langbein, L., Spring, H., Rogers, M.A., Praetzel, S., Schweizer, J. (2004). Hair keratins and hair follicle-specific epithelial keratins. *Methods in Cell Biology*, 78, 413-451.
2. Schmidt, A., Langbein, L., Rode, M., Praetzel, S., Zimbelmann, R., Franke, W.W. (1997). Plakophilins 1a and 1b: widespread nuclear proteins recruited in specific epithelial cells as desmosomal plaque components. *Cell Tiss. Res.* 290, 481-499.

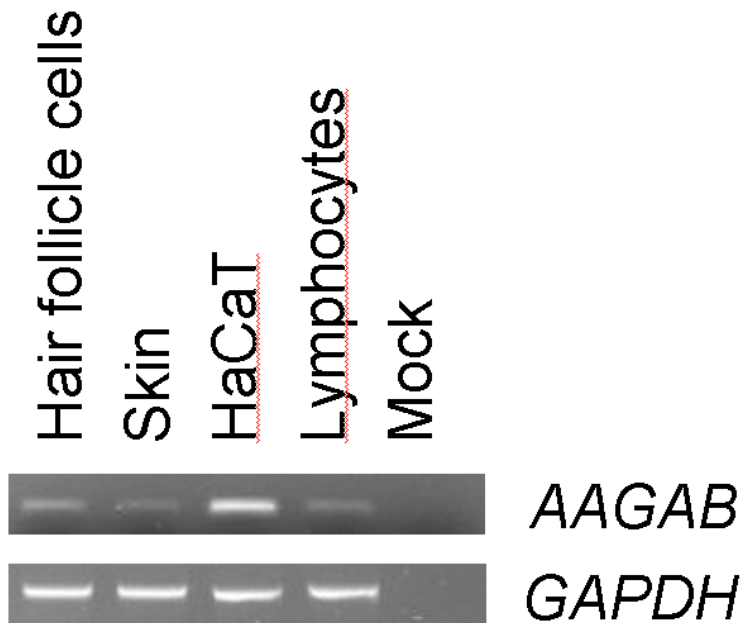


**Figure S1. HE Staining of Normal and Affected Footsole Epidermis**

(A) Normal footsole (control) skin. d, eccrine sweat gland duct.

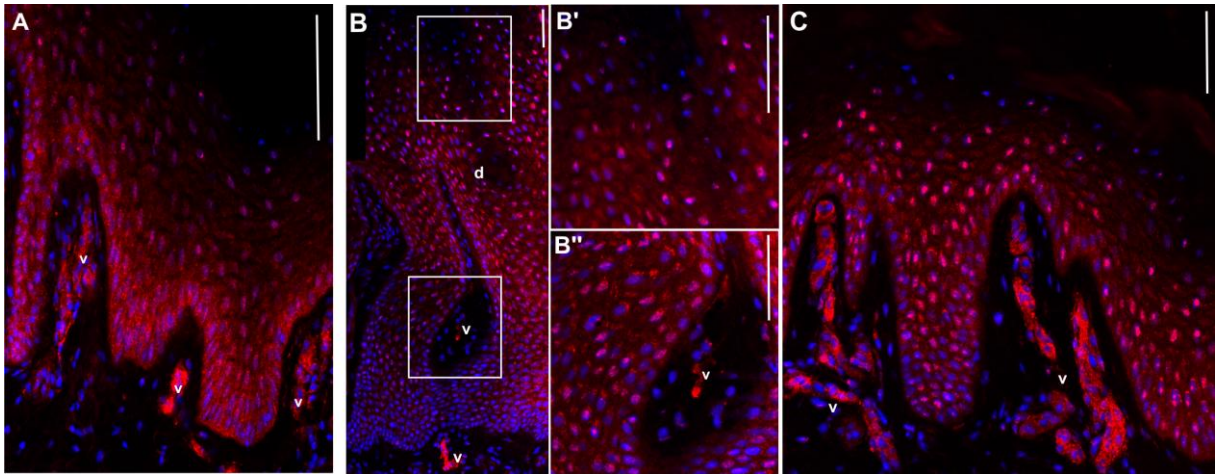
(B and C) Sole skin of an affected individual in the affected and (C) unaffected area. The affected skin (B) is much higher stratified including an enlarged stratum corneum when compared with the unaffected (C) area. The upper part of the stratum corneum was generally folded due to the extreme hyperkeratosis of pathogenetic affected skin sections.

Bars: 200  $\mu$ m.



**Figure S2. Expression Analysis in Human Tissues**

Expression of *AAGAB* and *GAPDH* in various normal human tissues and cells. RT-PCR analysis was performed from skin tissue, hair follicle cells, an immortalized keratinocyte cell line (HaCaT cells) and lymphocytes. The last lane—mock—is a negative (no template) control. *AAGAB* is expressed in all investigated tissues.



**Figure S3. Immunofluorescence Staining of AAGAB in Normal and Affected Skin**

(A) Normal control sole skin.

(B–B'' and C) Sole skin of an affected individual in the affected (B–B'') and unaffected (C) area. Upper and lower rectangles in (B) and (C) demarcate higher magnifications in B'/B'' respectively. v, dermal blood vessels; d, eccrine sweat gland ducts.

Bars: 100  $\mu$ m in A, B, and C; 15  $\mu$ m in B' and B''.

**Table S1. Disease Haplotypes of Families B and C in Chromosomal Region 15q22.31-15q23**

Name	Genetic Position	Physical Position	Pedigree B	Pedigree C	SNP
SNP15-67469128	73.1	67469128	C	A	[A/C]
SNP15-67469568	73.1	67469568	A	G	[A/G]
SNP15-67470208	73.1	67470208	T	T	[T/C]
SNP15-67471268	73.1	67471268	T	T	[T/C]
SNP15-67475083	73.1	67475083	G	G	[A/G]
SNP15-67476768	73.1	67476768	G	G	[A/G]
rs7173811	73.1	67479051	T	T	[T/C]
SNP15-67480480	73.1	67480480	C	C	[T/C]
SNP15-67480969	73.1	67480969	G	G	[A/G]
SNP15-67481514	73.1	67481514	C	C	[T/C]
rs8025774	73.1	67483276	C	C	[T/C]
rs8031440	73.1	67483979	C	C	[T/C]
rs8031627	73.1	67484119	C	C	[T/C]
SNP15-67485101	73.1	67485101	G	G	[A/G]
rs12595334	73.1	67485156	G	G	[A/G]
rs3743342	73.1	67485667	G	G	[A/G]
SNP15-67486078	73.1	67486078	C	C	[T/C]
rs12900401	73.1	67486590	G	G	[A/G]
rs3743343	73.1	67486775	C	C	[T/C]
rs1052488	73.1	67486847	G	G	[A/G]
SNP15-67492301	73.1	67492301	T	T	[T/C]
rs12476	73.1	67493726	A	A	[A/G]
SNP15-67494304	73.1	67494304	G	G	[T/G]
rs1050285	73.1	67494494	T	T	[T/C]
SNP15-67494922	73.1	67494922	T	T	[T/C]
SNP15-67498855	73.1	67498855	C	C	[T/C]
SNP15-67500330	73.1	67500330	T	T	[T/C]
SNP15-67501592	73.1	67501592	C	C	[T/C]
SNP15-67501782	73.1	67501782	G	G	[A/G]
SNP15-67502572	73.1	67502572	A	A	[A/G]
SNP15-67503637	73.1	67503637	G	G	[A/G]
rs7169664	73.1	67505066	G	G	[A/G]
rs7173826	73.1	67528374	A	A	[A/C]
rs11071943	73.1	67535996	A	A	[A/C]
rs12911370	73.1	67546321	G	G	[A/G]
SNP15-67548688	73.1	67548688	A	A	[A/G]
SNP15-67548840	73.1	67548840	C	C	[T/C]
SNP15-67549800	73.1	67549800	A	A	[A/C]
SNP15-67552064	73.1	67552064	G	G	[A/G]
SNP15-67555037	73.1	67555037	A	A	[A/G]
SNP15-67555487	73.1	67555487	C	C	[C/G]
SNP15-67555958	73.1	67555958	G	G	[A/G]
SNP15-67558010	73.1	67558010	G	G	[T/G]
SNP15-67561598	73.1	67561598	G	G	[A/G]
rs8029582	73.1	67575545	C	C	[A/C]
SNP15-67587368	73.1	67587368	T	T	[T/C]

<b>Name</b>	<b>Genetic Position</b>	<b>Physical Position</b>	<b>Pedigree B</b>	<b>Pedigree C</b>	<b>SNP</b>
SNP15-67599975	73.1	67599975	G	G	[A/G]
SNP15-67602464	73.1	67602464	C	C	[T/C]
rs11635391	73.1	67607900	A	A	[A/G]
rs7170004	73.1	67611732	T	T	[T/C]
SNP15-67614178	73.1	67614178	A	A	[A/C]
rs8032438	73.1	67615953	G	G	[A/G]
SNP15-67628649	73.1	67628649	G	G	[A/G]
rs956129	73.1	67629175	G	G	[A/G]
SNP15-67630587	73.1	67630587	C	C	[T/C]
SNP15-67632482	73.1	67632482	G	G	[A/G]
SNP15-67645243	73.1	67645243	T	T	[T/C]
rs10152973	73.1	67647218	A	A	[A/G]
SNP15-67648216	73.1	67648216	C	C	[T/C]
SNP15-67650155	73.1	67650155	T	T	[T/C]
rs4496076	73.1	67654369	G	G	[A/G]
SNP15-67663250	73.1	67663250	A	A	[A/G]
SNP15-67663284	73.1	67663284	A	A	[A/G]
SNP15-67664137	73.1	67664137	A	A	[A/G]
SNP15-67670535	73.1	67670535	C	C	[T/C]
SNP15-67680789	73.1	67680789	G	G	[T/G]
rs6494654	73.1	67681907	A	A	[A/G]
SNP15-67693890	73.1	67693890	A	A	[A/G]
rs950784	73.1	67697023	T	T	[T/C]
rs2286664	73.1	67701495	T	T	[T/C]
SNP15-67707134	73.1	67707134	C	C	[C/G]
SNP15-67710011	73.1	67710011	G	G	[A/G]
rs12148151	73.1	67715476	A	A	[A/G]
SNP15-67716497	73.1	67716497	T	T	[T/G]
rs4423371	73.1	67720855	G	G	[A/G]
SNP15-67725897	73.1	67725897	T	T	[T/C]
rs2899723	73.1	67736023	C	C	[T/C]
SNP15-67749871	73.1	67749871	A	A	[A/C]
SNP15-67753967	73.1	67753967	C	C	[T/C]
rs8038138	73.1	67756572	G	G	[A/G]
SNP15-67756899	73.1	67756899	A	A	[A/G]
SNP15-67760478	73.1	67760478	C	C	[T/C]
rs2899724	73.1	67760808	A	A	[A/G]
rs16950932	73.1	67761862	A	A	[A/G]
rs4776361	73.1	67762056	G	G	[A/G]
SNP15-67765450	73.1	67765450	A	A	[A/C]
rs12591605	73.1	67766364	G	G	[A/G]
SNP15-67770306	73.1	67770306	G	G	[T/G]
SNP15-67774033	73.1	67774033	G	G	[A/G]
SNP15-67784487	73.1	67784487	G	G	[A/G]
rs4278695	73.1	67787064	T	T	[T/C]
SNP15-67799130	73.1355	67799130	G	G	[A/G]
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SNP15-67804502	73.1584	67804502	G	G	[A/G]

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SNP15-67821056	73.2289	67821056	A	A	[A/G]
SNP15-67827705	73.2572	67827705	C	C	[T/C]
SNP15-67835370	73.2899	67835370	A	A	[A/G]
SNP15-67837009	73.2969	67837009	C	C	[T/C]
rs6494670	73.3204	67842526	G	G	[A/G]
SNP15-67845930	73.3349	67845930	C	C	[T/C]
SNP15-67850566	73.3547	67850566	G	G	[A/G]
rs11632574	73.3625	67852405	C	C,A	[A/C]
rs11632776	73.3635	67852645	T	T	[T/C]
SNP15-67863235	73.4086	67863235	T	T	[T/C]
rs11638507	73.4547	67874045	G	G	[A/G]
SNP15-67881637	73.487	67881637	G	G	[A/G]
rs10851775	73.5071	67886351	A	A	[A/G]
SNP15-67892792	73.5346	67892792	C	C	[T/C]
SNP15-67892994	73.5354	67892994	C	C	[T/C]
SNP15-67900883	73.5691	67900883	C	C	[T/C]
rs12593494	73.5718	67901532	A	A	[A/G]
rs2899726	73.5949	67906954	A	A	[A/G]
rs9672402	73.6056	67909473	G	G	[A/G]
rs11636945	73.6231	67913574	G	G	[A/G]
rs12905544	73.6738	67925452	A	A	[A/G]
SNP15-67929980	73.6931	67929980	G	G	[A/G]
rs11631474	73.7109	67934167	G	G	[A/G]
SNP15-67935745	73.7176	67935745	C	C	[T/C]
rs11638739	73.7341	67939612	A	A	[A/G]
SNP15-67942002	73.7443	67942002	G	G	[A/G]
rs4776371	73.7562	67944799	A	A	[A/G]
SNP15-67949040	73.7743	67949040	C	C	[T/C]
rs16951105	73.8452	67965694	A	A	[A/G]
SNP15-67969221	73.8603	67969221	T	T	[T/G]
SNP15-67972908	73.876	67972908	T	T	[T/C]
rs11637445	73.9557	67991606	G	G	[T/G]
SNP15-67993166	73.9623	67993166	A	A	[A/G]
rs11632828	73.9695	67994849	G	G	[A/G]
SNP15-68002488	74.002	68002488	C	C	[T/C]
SNP15-68012403	74.0443	68012403	A	A	[T/C]
SNP15-68013079	74.0472	68013079	G	A,G	[A/G]
rs997295	74.0611	68016343	A	A	[A/C]
SNP15-68016955	74.0636	68016955	A	A	[A/C]
rs3784700	74.0671	68017752	A	A	[A/G]
rs10518740	74.0729	68019118	C	C	[T/C]
SNP15-68019958	74.0765	68019958	A	A	[A/G]
rs3784704	74.115	68028989	A	A	[A/G]
rs12905397	74.1369	68034150	G	G	[A/G]
rs12593813	74.1484	68036852	G	G	[A/G]



<b>Name</b>	<b>Genetic Position</b>	<b>Physical Position</b>	<b>Pedigree B</b>	<b>Pedigree C</b>	<b>SNP</b>
rs11635424	74.1515	68037578	C	C	[T/C]
rs7177848	74.1674	68041299	C	C	[T/C]
rs16951227	74.1765	68043433	A	A	[A/G]
SNP15-68045430	74.185	68045430	G	G	[A/G]
SNP15-68046528	74.1897	68046528	G	G	[A/G]
rs8042507	74.2001	68048974	A	A	[A/C]
rs7183639	74.2078	68050777	G	G	[A/G]
SNP15-68051608	74.2113	68051608	G	G	[A/G]
rs884202	74.2232	68054388	A	A	[A/G]
SNP15-68055570	74.2282	68055570	G	G	[A/G]
SNP15-68056480	74.2321	68056480	G	G	[T/G]
rs11071959	74.2424	68058903	G	G	[A/G]
rs745212	74.2478	68060179	A	A	[A/G]
rs10518744	74.2598	68062975	C	C	[T/C]
SNP15-68063546	74.2622	68063546	G	G	[A/G]
rs4489954	74.2985	68072075	C	C	[A/C]
rs3784709	74.2994	68072275	G	G	[A/G]
rs3784711	74.3005	68072527	C	C	[T/C]
rs4261478	74.3071	68074084	C	C	[T/C]
SNP15-68075975	74.3152	68075975	C	C	[T/C]
rs12902904	74.3176	68076533	T	T	[T/C]
SNP15-68079358	74.3296	68079358	G	G	[A/G]
SNP15-68083436	74.347	68083436	T	T	[T/G]
rs3784716	74.3513	68084450	A	A	[A/C]
rs2241423	74.3615	68086838	C	C	[T/C]
rs16951304	74.3733	68089618	A	A	[A/G]
rs11635396	74.3767	68090423	A	A	[A/G]
rs2127162	74.3784	68090824	T	T	[T/G]
rs1026732	74.3966	68095085	C	C	[T/C]
rs13329413	74.4053	68097126	C	C	[T/C]
SNP15-68098936	74.413	68098936	G	G	[A/G]
SNP15-68100330	74.4189	68100330	G	G	[T/G]
rs16951319	74.433	68103632	G	G	[A/G]
SNP15-68103695	74.4333	68103695	C	C	[T/C]
rs7180312	74.4399	68105256	G	G	[A/G]
SNP15-68105878	74.4426	68105878	T	T	[T/C]
SNP15-68109907	74.4597	68109907	T	T	[T/C]
rs2061845	74.4698	68112274	G	G	[A/G]
rs4776982	74.4814	68114974	A	A	[A/G]
SNP15-68116400	74.4874	68116400	G	G	[T/G]
SNP15-68123582	74.518	68123582	G	G	[A/G]
SNP15-68124665	74.5226	68124665	C	C	[A/C]
rs3803522	74.5312	68126675	G	G	[A/G]
SNP15-68127773	74.5359	68127773	G	G	[A/G]
SNP15-68129127	74.5417	68129127	G	G	[T/G]
SNP15-68129326	74.5425	68129326	T	T	[T/G]
rs7497457	74.557	68132723	T	T	[T/G]
SNP15-68133617	74.5608	68133617	T	T	[T/C]

Name	Genetic Position	Physical Position	Pedigree B	Pedigree C	SNP
rs12907778	74.5668	68135027	A	A	[A/G]
rs16951344	74.5788	68137843	A	A	[A/C]
rs4776991	74.5887	68140162	A	A	[A/G]
SNP15-68140610	74.5906	68140610	A	A	[A/G]
SNP15-68141566	74.5947	68141566	C	C	[A/C]
SNP15-68143288	74.602	68143288	C	C	[T/C]
rs11637396	74.6065	68144345	C	C	[T/C]
SNP15-68144825	74.6085	68144825	T	T	[T/C]
rs12905141	74.6167	68146731	C	C	[T/C]
rs12592776	74.6239	68148432	G	G	[T/G]
rs1001870	74.6368	68151448	T	T	[T/C]
SNP15-68152190	74.6399	68152190	G	A,G	[A/G]
SNP15-68152198	74.64	68152198	C	A,C	[T/C]
SNP15-68155644	74.6547	68155644	C	C	[T/C]
rs4776381	74.6549	68155710	A	A	[A/G]
rs1982647	74.6564	68156055	A	A	[A/G]
rs4306451	74.6642	68157881	G	G	[A/G]
SNP15-68162131	74.6823	68162131	T	T	[T/C]
SNP15-68166930	74.7027	68166930	A	A	[A/G]
rs8037951	74.7378	68175157	A	A	[A/C]
SNP15-68176397	74.7431	68176397	C	C	[T/C]
SNP15-68177714	74.7487	68177714	C	A,C	[T/C]
SNP15-68180081	74.7588	68180081	C	C	[T/C]
SNP15-68180471	74.7605	68180471	G	G	[A/G]
rs1996120	74.765	68181534	A	A	[A/G]
SNP15-68183005	74.7713	68183005	T	T	[T/C]
SNP15-68186641	74.7867	68186641	G	G	[A/G]
rs8032153	74.8225	68195035	A	A	[A/G]
SNP15-68198140	74.8358	68198140	G	G	[A/G]
rs430610	74.8378	68198622	T	T	[T/C]
SNP15-68198741	74.8383	68198741	C	C	[T/C]
SNP15-68202649	74.855	68202649	G	T,G	[T/G]
SNP15-68204357	74.8622	68204357	C	C	[A/C]
SNP15-68207015	74.8736	68207015	T	T	[T/C]
SNP15-68207020	74.8736	68207020	A	G,A	[A/G]
SNP15-68211991	74.8948	68211991	C	C	[T/C]
rs338369	74.9088	68215300	C	C	[A/C]
rs338372	74.9185	68217562	G	G	[A/G]
SNP15-68217809	74.9196	68217809	C	C	[T/C]
rs11633495	74.9243	68218922	G	A,G	[A/G]
rs16951479	74.9245	68218974	C	A,C	[A/C]
SNP15-68219974	74.9288	68219974	T	T	[T/C]
SNP15-68220072	74.9292	68220072	C	C	[A/C]
rs396174	74.9335	68221078	A	A	[A/G]
rs2448134	74.9419	68223040	C	C	[T/C]
SNP15-68225244	74.9513	68225244	C	T,C	[T/C]
SNP15-68227960	74.9628	68227960	A	A	[A/G]
SNP15-68227985	74.9629	68227985	T	T	[T/C]

Name	Genetic Position	Physical Position	Pedigree B	Pedigree C	SNP
rs338350	74.9675	68229052	C	C	[T/C]
SNP15-68232499	74.9821	68232499	T	T	[T/G]
rs1564277	74.988	68233869	C	C	[T/C]
rs11636387	74.992	68234818	T	T	[T/C]
SNP15-68235621	74.9955	68235621	G	G	[A/G]
rs44410	74.9956	68235646	A	A	[A/G]
SNP15-68236286	74.9983	68236286	T	T	[T/C]
SNP15-68240825	75.0176	68240825	T	G,T	[T/G]
rs12439223	75.0207	68241646	T	T	[T/C]
SNP15-68242193	75.0221	68242193	C	C	[T/C]
rs8036635	75.0308	68245488	A	C,A	[A/C]
SNP15-68245930	75.032	68245930	A	T,A	[A/T]
rs12902929	75.0356	68247308	C	A,C	[A/C]
SNP15-68249135	75.0404	68249135	G	G	[A/G]
SNP15-68250076	75.0429	68250076	C	C	[A/C]
SNP15-68253238	75.0512	68253238	A	G,A	[A/G]
SNP15-68254156	75.0536	68254156	A	A	[A/G]
SNP15-68254367	75.0542	68254367	T	G,T	[T/G]
rs16951524	75.0546	68254541	C	T,C	[T/C]
SNP15-68254591	75.0548	68254591	A	A,G	[A/G]
rs338389	75.069	68260008	G	A,G	[A/G]
rs10851782	75.0823	68265049	A	A	[A/G]
rs338344	75.0963	68270380	T	T	[T/C]
rs338342	75.1007	68272052	G	G	[A/G]
SNP15-68273986	75.1058	68273986	T	T,C	[T/C]
SNP15-68274789	75.1079	68274789	T	C,T	[T/C]
SNP15-68276343	75.112	68276343	C	C	[T/C]
rs8037902	75.1234	68280703	A	A	[A/G]
SNP15-68281008	75.1242	68281008	G	G	[A/G]
SNP15-68282153	75.1272	68282153	A	A	[T/A]
SNP15-68285356	75.1357	68285356	A	A	[A/G]
SNP15-68285578	75.1362	68285578	G	G	[A/G]
SNP15-68288459	75.1438	68288459	C	C	[T/C]
rs7163804	75.1504	68290946	A	A	[A/C]
rs6494709	75.1584	68294010	G	G	[A/G]
rs1532113	75.16	68294598	C	C	[T/C]
SNP15-68296888	75.166	68296888	A	A	[A/G]
rs16951552	75.1798	68302144	C	C	[T/C]
SNP15-68305370	75.1883	68305370	A	A	[A/G]
rs11071974	75.1911	68306447	G	G	[A/G]
SNP15-68307970	75.1951	68307970	A	A	[A/G]
rs4777016	75.2134	68314919	C	C	[T/C]
SNP15-68317407	75.22	68317407	A	A	[A/G]
rs16951597	75.2321	68322007	G	G	[A/G]
rs12593922	75.2424	68325938	A	A	[A/G]
SNP15-68329025	75.2505	68329025	A	A	[A/G]
SNP15-68330375	75.2541	68330375	G	G	[A/G]
SNP15-68338706	75.276	68338706	G	A,G	[A/G]

Name	Genetic Position	Physical Position	Pedigree B	Pedigree C	SNP
rs1489598	75.3054	68349893	G	G	[A/G]
SNP15-68351048	75.3084	68351048	C	C	[A/C]
SNP15-68373638	75.3678	68373638	T	T	[T/C]
rs1489595	75.377	68377126	T	T	[T/C]
SNP15-68381444	75.3884	68381444	T	T	[T/C]
SNP15-68384227	75.3957	68384227	C	C	[A/C]
rs8025474	75.4017	68386523	G	A,G	[A/G]
SNP15-68392332	75.417	68392332	G	G	[A/G]
rs4306484	75.4276	68396372	G	G	[A/G]
rs11856619	75.4486	68404362	C	C	[T/C]
SNP15-68404831	75.4499	68404831	A	G,A	[A/G]
SNP15-68417093	75.4821	68417093	T	T	[T/C]
SNP15-68425635	75.5046	68425635	A	A	[A/G]
SNP15-68432140	75.5217	68432140	G	G	[A/G]
rs8035890	75.5348	68437102	T	T	[T/G]
rs4777022	75.5425	68440028	T	T	[T/C]
SNP15-68440466	75.5436	68440466	G	G	[A/G]
SNP15-68440606	75.544	68440606	C	T,C	[T/C]
SNP15-68445679	75.5573	68445679	G	G	[A/G]
rs10152713	75.6262	68471891	C	C	[T/C]
SNP15-68475424	75.6355	68475424	T	C,T	[T/C]
rs1049493	75.6499	68480887	A	A	[A/G]
SNP15-68486097	75.6636	68486097	A	A	[A/G]
rs11071986	75.6792	68492028	A	A	[A/G]
rs11071991	75.695	68498052	C	C	[T/C]
rs8028241	75.6984	68499321	G	G	[A/G]
SNP15-68499661	75.6993	68499661	T	T	[T/C]
rs1400432	75.707	68502604	A	A	[A/G]
SNP15-68503216	75.7086	68503216	A	A	[A/G]
rs17317291	75.7523	68519814	G	G	[T/G]
rs2169215	75.8024	68538856	T	T	[T/C]
rs8038170	75.8136	68543128	T	T	[T/C]
rs1554223	75.8201	68545603	A	A	[A/G]
SNP15-68545798	75.8206	68545798	A	A	[A/G]
SNP15-68546156	75.8216	68546156	A	A	[A/G]
rs12902478	75.8259	68547828	A	A	[A/G]
SNP15-68553914	75.842	68553914	G	T,G	[T/G]
rs2280218	75.8764	68567012	A	A	[A/C]
SNP15-68567752	75.8784	68567752	C	C	[T/C]
rs16951723	75.8843	68570011	C	T,C	[T/C]
SNP15-68570158	75.8847	68570158	T	T,G	[T/G]
SNP15-68571164	75.8873	68571164	C	T,C	[T/C]
rs10152450	75.8882	68571479	A	A,C	[A/C]
SNP15-68578329	75.9062	68578329	T	T,G	[T/G]
rs12909277	75.913	68580912	G	A,G	[A/G]
rs10444850	75.9341	68588935	C	T,C	[T/C]
SNP15-68594522	75.9488	68594522	C	T,C	[T/C]
SNP15-68594861	75.9497	68594861	G	A,G	[A/G]

Name	Genetic Position	Physical Position	Pedigree B	Pedigree C	SNP
SNP15-68595801	75.9521	68595801	C	T,C	[T/C]
rs7174755	75.9556	68597127	T	T,C	[T/C]
rs898590	75.9741	68604175	C	T,C	[T/C]
SNP15-68605067	75.9765	68605067	A	A	[A/C]
rs4777035	75.9768	68605169	A	A,G	[A/G]
rs1125721	75.9807	68606656	G	G	[A/G]
SNP15-68608187	75.9847	68608187	A	A	[A/T]
SNP15-68611426	75.9932	68611426	G	G	[T/C]
SNP15-68611489	75.9934	68611489	C	C	[G/C]
SNP15-68612890	75.9971	68612890	A	A	[A/C]
SNP15-68613778	75.9994	68613778	G	G	[T/G]
rs3736493	75.9994	68613787	T	T	[T/C]
SNP15-68614895	76.0023	68614895	A	A	[A/G]
SNP15-68615422	76.0037	68615422	A	A	[A/G]
rs2414997	76.004	68615519	T	T	[T/C]
rs1516869	76.0049	68615873	G	G	[A/G]
SNP15-68616678	76.007	68616678	T	T	[T/C]
rs4777037	76.0161	68620136	C	C	[A/C]
rs2292745	76.0271	68624290	T	T	[T/C]
SNP15-68624396	76.0273	68624396	C	C	[A/C]
SNP15-68625175	76.0294	68625175	A	A	[A/G]
SNP15-68625456	76.0301	68625456	T	T	[T/G]
rs964691	76.033	68626542	G	G	[A/G]
rs2306024	76.0369	68628049	A	A	[A/C]
rs2306023	76.0372	68628143	T	T	[T/C]
rs898586	76.0384	68628601	C	C	[T/C]
SNP15-68628636	76.0385	68628636	A	A	[A/G]
rs898585	76.0394	68628999	G	G	[A/G]
SNP15-68629763	76.0414	68629763	C	C	[T/C]
SNP15-68630204	76.0426	68630204	T	T	[T/C]
rs7179347	76.0444	68630870	G	G	[A/G]
rs2125998	76.0466	68631740	G	G	[A/G]
rs7169899	76.0514	68633534	G	G	[A/G]
rs8035990	76.052	68633773	C	C	[T/C]
SNP15-68634536	76.054	68634536	G	G	[A/G]
rs898582	76.0567	68635575	C	C	[T/C]
rs1563894	76.0573	68635775	A	A	[A/G]
SNP15-68636432	76.059	68636432	A	A	[A/G]
rs3784344	76.06	68636798	T	T	[T/C]
SNP15-68636849	76.0601	68636849	C	C	[T/C]
SNP15-68638084	76.0633	68638084	A	A	[A/G]
SNP15-68639206	76.0663	68639206	A	A	[A/G]
SNP15-68639385	76.0667	68639385	T	T	[T/C]
SNP15-68639606	76.0673	68639606	A	A	[A/G]
rs8029838	76.0692	68640333	C	C	[T/C]
SNP15-68640452	76.0695	68640452	A	A	[A/G]
SNP15-68641741	76.073	68641741	T	T	[T/C]
rs3784350	76.0738	68642047	G	G	[A/G]

Name	Genetic Position	Physical Position	Pedigree B	Pedigree C	SNP
rs3784351	76.0742	68642220	C	C,T	[T/C]
SNP15-68647590	76.0883	68647590	A	A	[A/G]
rs12907890	76.0964	68649587	T	T	[T/C]
SNP15-68651641	76.1062	68651641	A	A	[A/G]
SNP15-68656035	76.127	68656035	T	T	[T/G]
rs8040014	76.1276	68656162	T	T	[T/C]
rs2306012	76.1314	68656960	G	G	[A/G]
SNP15-68659450	76.1432	68659450	T	T	[T/C]
rs2415009	76.1463	68660104	T	T	[T/C]
SNP15-68660703	76.1492	68660703	G	G	[A/G]
SNP15-68660823	76.1497	68660823	G	G	[A/G]
SNP15-68661281	76.1519	68661281	C	T,C	[T/C]
rs1460644	76.1557	68662081	T	T	[T/C]
rs16951961	76.1599	68662976	C	C	[T/C]
rs11072011	76.1614	68663283	G	G	[A/G]
rs12595773	76.1696	68665017	C	C	[T/C]
rs967565	76.1722	68665570	C	C	[T/C]
SNP15-68666643	76.1773	68666643	T	T	[T/C]
rs7183704	76.1876	68668816	T	T	[T/G]
rs1460645	76.1896	68669239	C	C	[T/C]
rs11858758	76.1955	68670469	G	G	[A/G]
SNP15-68671867	76.2021	68671867	T	T	[T/C]
SNP15-68673835	76.2114	68673835	T	T	[T/C]
SNP15-68675236	76.2181	68675236	G	G	[A/G]
rs4777048	76.2292	68677576	T	T	[T/G]
SNP15-68678731	76.2346	68678731	G	G	[A/G]
rs1075991	76.2368	68679181	T	T	[T/C]
SNP15-68680258	76.2404	68680258	A	A	[A/G]
SNP15-68680390	76.2405	68680390	G	G	[A/G]
SNP15-68681554	76.2416	68681554	A	A	[A/G]
rs11638575	76.2424	68682309	T	T	[T/G]
SNP15-68682612	76.2427	68682612	G	G	[A/G]
rs7173365	76.245	68684970	C	C	[A/C]
SNP15-68685340	76.2453	68685340	T	T	[T/A]
SNP15-68686333	76.2463	68686333	G	G	[A/G]
SNP15-68686852	76.2468	68686852	G	G	[A/G]
rs11629460	76.255	68695349	C	C	[T/C]
rs11630897	76.2562	68696621	G	G	[A/G]
rs11857615	76.2567	68697093	T	T	[T/C]
rs12050670	76.2574	68697797	A	A	[A/C]
rs9972560	76.2577	68698138	C	C	[A/C]
rs11072020	76.2578	68698192	C	C	[T/C]
rs7162189	76.2583	68698803	A	A	[A/G]
rs11635082	76.2587	68699163	C	C	[T/C]
rs17320635	76.2611	68701651	A	A	[A/G]
rs7176145	76.2627	68703268	T	T	[T/G]
SNP15-68704101	76.2635	68704101	A	A	[A/G]
rs898577	76.265	68705697	C	C	[T/C]

Name	Genetic Position	Physical Position	Pedigree B	Pedigree C	SNP
SNP15-68706284	76.2656	68706284	A	A	[A/G]
SNP15-68708179	76.2674	68708179	T	T	[T/C]
SNP15-68708629	76.2679	68708629	A	A	[A/G]
SNP15-68708723	76.2679	68708723	C	C	[T/C]
rs7169854	76.2688	68709584	G	G	[A/G]
SNP15-68711017	76.2702	68711017	G	G	[A/G]
rs8024010	76.2706	68711470	T	T	[T/G]
SNP15-68711518	76.2707	68711518	G	G	[A/G]
SNP15-68711649	76.2708	68711649	G	G	[A/G]
SNP15-68712107	76.2712	68712107	G	G	[A/G]
SNP15-68712460	76.2716	68712460	G	G	[A/G]
SNP15-68713065	76.2722	68713065	G	G	[A/G]
SNP15-68713617	76.2727	68713617	G	G	[A/G]
SNP15-68713635	76.2727	68713635	G	G	[A/G]
SNP15-68714245	76.2733	68714245	C	C	[T/C]
SNP15-68715486	76.2745	68715486	T	T	[T/C]
SNP15-68716567	76.2755	68716567	G	G	[A/G]
rs8036698	76.2758	68716776	T	T	[T/C]
SNP15-68717979	76.2769	68717979	C	C	[T/C]
rs16952059	76.2769	68718000	A	A	[A/G]
rs16952065	76.2779	68718987	C	C	[T/C]
SNP15-68721538	76.2804	68721538	G	G	[T/G]
SNP15-68722165	76.281	68722165	T	T	[T/C]
SNP15-68722374	76.2812	68722374	T	T	[T/C]
SNP15-68723348	76.2821	68723348	C	T	[T/C]
rs895135	76.2825	68723712	T	G	[T/G]
SNP15-68728094	76.2867	68728094	T	C	[T/C]
SNP15-68728632	76.2872	68728632	G	T	[T/G]

The markers within *AAGAB* are displayed in green. All alleles where the haplotype of family B differs from that of family C are displayed in purple. At ambiguous loci which can't be phased using the available information, both alleles are indicated. The nucleotide and the physical position of the markers refer to the human genome assembly hg19. Markers which do not have an rs-number are named according to Illumina's chip annotation.