

Supplementary Material

Materials and Methods

Immunological and other laboratory analyses were done as routine tests in the institutional labs except otherwise indicated. In brief, for FACS analyses whole blood (50 μ l) of patients and healthy controls was stained with optimal concentrations of the indicated directly conjugated monoclonal antibodies at room temperature for 20 minutes. Subsequently, red blood cells were removed with ADG-lysis solution (An der Grub, Vienna, Austria) and 1×10^4 cells in the lymphogate were acquired, gated on CD19⁺ cells and further analyzed for IgD and CD27 expression as described before (see Refs. 16, 17 of the main document). CGH array analyses were done as described earlier (see Ref. 14 of the main document). For FISH analyses mix 7 and mix 14 of the Vysis ToTelVysion Multi-Color FISH Probe Kit (Abbott Diagnostics, Vienna, Austria) were used. Mix 7 of the kit contains probes TelVysion 7p SpectrumGreen, TelVysion 7q SpectrumOrange, TelVysion 14q SpectrumGreen and SpectrumOrange and LSI TCR 14q11.2 SpectrumAqua. Mix14 contains probes TelVysion 19p SpectrumGreen, TelVysion 19q SpectrumOrange and LSI 19p13 SpectrumAqua. Metaphase preparation was carried out by standard methods. Briefly, cells were cultured in LymphoGrow medium (CytoGen, Sinn, Germany) containing phytohaemagglutinin as mitogen. After fixation, metaphases were dropped onto slides and then dried at 60°C overnight. One slide was used for G-banding. For FISH analyses ToTelVysion probes were used after manufactures instruction, however, metaphase slides were hybridised without pretreatment.

Supplementary Table 1. Protein-encoding genes within the duplicated interval on 19p13 for patient 1 and 2 as indicated by array CGH analysis results annotated using Human Genome Build GRCh37/hg19.

Patient	Gene Name	Description
1	<i>OR4F17</i>	olfactory receptor
1	<i>PPAP2C</i>	phosphatidic acid phosphatase type 2C
1	<i>MIER2</i>	mesoderm induction early response 1
1+2	<i>THEG</i>	theg spermatid protein
1+2	<i>C2CD4C</i>	C2 calcium-dependent domain containing 4C
1+2	<i>SHC2</i>	SHC (Src homology 2 domain containing) transforming protein 2
1+2	<i>ODF3L2</i>	outer dense fiber of sperm tails 3-like 2
1+2	<i>MADCAM1</i>	mucosal vascular addressin cell adhesion molecule 1
1+2	<i>TPGS1</i>	tubulin polyglutamylase complex subunit 1
1+2	<i>CDC34</i>	cell division cycle 34
1+2	<i>GZMM</i>	granzyme M (lymphocyte met-ase 1)
1+2	<i>BSG</i>	basigin (Ok blood group) hyperpolarization activated cyclic nucleotide-gated potassium channel 2
1+2	<i>HCN2</i>	channel 2
1+2	<i>POLRMT</i>	polymerase (RNA) mitochondrial (DNA directed)
1+2	<i>FGF22</i>	fibroblast growth factor 22
1+2	<i>RNF126</i>	ring finger protein 126
1+2	<i>FSTL3</i>	follistatin-like 3 (secreted glycoprotein)
1+2	<i>PRSS57</i>	protease
1+2	<i>PALM</i>	paralemmin
1+2	<i>C19orf21</i>	chromosome 19 open reading frame 21
1+2	<i>PTBPI</i>	polypyrimidine tract binding protein 1
1+2	<i>LPPR3</i>	hsa-mir-3187
1+2	<i>AZU1</i>	azurocidin 1
1+2	<i>PRTN3</i>	proteinase 3
1+2	<i>ELANE</i>	elastase
1+2	<i>CFD</i>	complement factor D (adipsin)
1+2	<i>MED16</i>	mediator complex subunit 16
1+2	<i>R3HDM4</i>	R3H domain containing 4
1+2	<i>KISS1R</i>	KISS1 receptor
1+2	<i>ARID3A</i>	AT rich interactive domain 3A (BRIGHT-like)
1+2	<i>WDR18</i>	WD repeat domain 18
1+2	<i>AC004528.1</i>	Uncharacterized protein
1+2	<i>GRIN3B</i>	glutamate receptor
1+2	<i>TMEM259</i>	transmembrane protein 259
1+2	<i>CNN2</i>	calponin 2
1+2	<i>ABCA7</i>	ATP-binding cassette
1+2	<i>HMHA1</i>	histocompatibility (minor) HA-1
1+2	<i>POLR2E</i>	polymerase (RNA) II (DNA directed) polypeptide E
1+2	<i>GPX4</i>	glutathione peroxidase 4
1+2	<i>SBNO2</i>	strawberry notch homolog 2 (Drosophila)

1+2	<i>STK11</i>	serine/threonine kinase 11
1+2	<i>C19orf26</i>	chromosome 19 open reading frame 26
1+2	<i>ATP5D</i>	ATP synthase
1+2	<i>MIDN</i>	midnolin
1+2	<i>CIRBP</i>	cold inducible RNA binding protein
1+2	<i>C19orf24</i>	chromosome 19 open reading frame 24
1+2	<i>MUM1</i>	melanoma associated antigen (mutated) 1
1+2	<i>EFNA2</i>	ephrin-A2
1+2	<i>NDUFS7</i>	NADH dehydrogenase (ubiquinone) Fe-S protein 7
1+2	<i>GAMT</i>	guanidinoacetate N-methyltransferase
1+2	<i>DAZAP1</i>	DAZ associated protein 1
1+2	<i>RPS15</i>	ribosomal protein S15
1+2	<i>AC027307.3</i>	Uncharacterized protein
1+2	<i>APC2</i>	adenomatosis polyposis coli 2
1+2	<i>C19orf25</i>	chromosome 19 open reading frame 25
1+2	<i>PCSK4</i>	proprotein convertase subtilisin/kexin type 4
1+2	<i>REEP6</i>	receptor accessory protein 6
1+2	<i>ADAMTSL5</i>	ADAMTS-like 5
1+2	<i>PLK5</i>	polo-like kinase 5
1+2	<i>MEX3D</i>	mex-3 homolog D (<i>C. elegans</i>)
1+2	<i>MBD3</i>	methyl-CpG binding domain protein 3
1+2	<i>UQCRI1</i>	cytochrome b-c1 complex subunit 10
1+2	<i>TCF3</i>	transcription factor 3
1+2	<i>ONECUT3</i>	one cut homeobox 3
1+2	<i>ATP8B3</i>	ATPase
1+2	<i>REXO1</i>	REX1
1+2	<i>KLF16</i>	Kruppel-like factor 16
1+2	<i>ABHD17A</i>	abhydrolase domain containing 17A
1+2	<i>SCAMP4</i>	secretory carrier membrane protein 4
1+2	<i>ADAT3</i>	adenosine deaminase
1+2	<i>CSNK1G2</i>	casein kinase 1
1+2	<i>BTBD2</i>	BTB (POZ) domain containing 2
1+2	<i>MKNK2</i>	MAP kinase interacting serine/threonine kinase 2
1+2	<i>MOB3A</i>	MOB kinase activator 3A
1+2	<i>IZUMO4</i>	IZUMO family member 4
1+2	<i>AP3D1</i>	adaptor-related protein complex 3
1+2	<i>DOT1L</i>	DOT1-like
1+2	<i>PLEKHJ1</i>	pleckstrin homology domain containing
1+2	<i>SF3A2</i>	splicing factor 3a
1+2	<i>AMH</i>	anti-Mullerian hormone
1+2	<i>JSRPI</i>	junctional sarcoplasmic reticulum protein 1
1+2	<i>OAZ1</i>	ornithine decarboxylase antizyme 1
1+2	<i>C19orf35</i>	chromosome 19 open reading frame 35
1+2	<i>LINGO3</i>	leucine rich repeat and Ig domain containing 3
1+2	<i>LSM7</i>	LSM7 homolog
1+2	<i>TMPRSS9</i>	transmembrane protease
1+2	<i>TIMM13</i>	translocase of inner mitochondrial membrane 13 homolog (yeast)

1+2	LMNB2	lamin B2
1+2	GADD45B	growth arrest and DNA-damage-inducible
1+2	GNG7	guanine nucleotide binding protein (G protein)
1+2	DIRAS1	DIRAS family
1+2	AC006538.4	Uncharacterized protein
1+2	SLC39A3	solute carrier family 39 (zinc transporter)
1+2	SGTA	small glutamine-rich tetratricopeptide repeat (TPR)-containing
1+2	THOP1	thimet oligopeptidase 1
1+2	ZNF554	zinc finger protein 554
1+2	ZNF555	zinc finger protein 555
1+2	ZNF556	zinc finger protein 556
1+2	ZNF57	zinc finger protein 57
1+2	ZNF77	zinc finger protein 77
1+2	TLE6	transducin-like enhancer of split 6 (E(sp1) homolog
1+2	TLE2	transducin-like enhancer of split 2 (E(sp1) homolog
1+2	AES	amino-terminal enhancer of split
1+2	GNA11	guanine nucleotide binding protein (G protein)
1+2	GNA15	guanine nucleotide binding protein (G protein)
1+2	SIPR4	sphingosine-1-phosphate receptor 4
1+2	NCLN	nicalin
1+2	CELF5	CUGBP
1+2	NFIC	nuclear factor I/C (CCAAT-binding transcription factor)
1+2	C19orf77	chromosome 19 open reading frame 77
1+2	DOHH	deoxyhypusine hydroxylase/monooxygenase
1+2	FZRI	fizzy/cell division cycle 20 related 1 (Drosophila)
1+2	MFSD12	major facilitator superfamily domain containing 12
1+2	C19orf71	chromosome 19 open reading frame 71
1+2	HMG20B	high mobility group 20B
1+2	GIPC3	GIPC PDZ domain containing family
1+2	TBXA2R	thromboxane A2 receptor
1+2	CACTIN	cactin
1+2	PIP5K1C	phosphatidylinositol-4-phosphate 5-kinase
1+2	TJP3	tight junction protein 3
1+2	APBA3	amyloid beta (A4) precursor protein-binding
1+2	MRPL54	mitochondrial ribosomal protein L54
1+2	RAX2	retina and anterior neural fold homeobox 2
1+2	MATK	megakaryocyte-associated tyrosine kinase
1+2	ZFR2	zinc finger RNA binding protein 2
1+2	ATCAY	ataxia
1+2	NMRK2	nicotinamide riboside kinase 2
1+2	DAPK3	death-associated protein kinase 3
1+2	EEF2	eukaryotic translation elongation factor 2
1+2	PIAS4	protein inhibitor of activated STAT
1+2	ZBTB7A	zinc finger and BTB domain containing 7A
1+2	MAP2K2	mitogen-activated protein kinase kinase 2
1+2	CREB3L3	cAMP responsive element binding protein 3-like 3
1+2	SIRT6	sirtuin 6

1+2	ANKRD24	ankyrin repeat domain 24
1+2	EBI3	Epstein-Barr virus induced 3
1+2	CCDC94	coiled-coil domain containing 94
1+2	SHD	Src homology 2 domain containing transforming protein D
1+2	TMIGD2	transmembrane and immunoglobulin domain containing 2
1+2	FSD1	fibronectin type III and SPRY domain containing 1
1+2	STAP2	signal transducing adaptor family member 2
1+2	MPND	MPN domain containing
1+2	SH3GL1	SH3-domain GRB2-like 1
1+2	CHAF1A	chromatin assembly factor 1
1+2	UBXN6	UBX domain protein 6
1+2	HDGFRP2	Hepatoma-derived growth factor-related protein 2
1+2	PLIN4	perilipin 4
1+2	PLIN5	perilipin 5
	CTB-	
1+2	50L17.14	Uncharacterized protein
1+2	LRG1	leucine-rich alpha-2-glycoprotein 1
1+2	SEMA6B	sema domain
1+2	TNFAIP8L1	tumor necrosis factor
1+2	C19orf10	chromosome 19 open reading frame 10
1+2	DPP9	dipeptidyl-peptidase 9
1+2	FEM1A	fem-1 homolog a (C. elegans)
1+2	TICAM1	toll-like receptor adaptor molecule 1
1+2	PLIN3	perilipin 3
1+2	ARRDC5	arrestin domain containing 5
1+2	KDM4B	lysine (K)-specific demethylase 4B
1+2	PTPRS	protein tyrosine phosphatase
1+2	ZNRF4	zinc and ring finger 4
1+2	SAFB2	scaffold attachment factor B2
1+2	SAFB	scaffold attachment factor B
1+2	RPL36	ribosomal protein L36
1+2	C19orf70	chromosome 19 open reading frame 70
1+2	HSD11B1L	hydroxysteroid (11-beta) dehydrogenase 1-like
1+2	LONP1	lon peptidase 1
1+2	CATSPERD	catsper channel auxiliary subunit delta
1+2	PRR22	proline rich 22
1+2	CTB-5409.9	Uncharacterized protein
1+2	DUS3L	dihydrouridine synthase 3-like (S. cerevisiae)
1+2	NRTN	neurturin
1+2	FUT6	fucosyltransferase 6 (alpha (1
1+2	FUT3	fucosyltransferase 3 (galactoside 3(4)-L-fucosyltransferase
1+2	FUT5	fucosyltransferase 5 (alpha (1
1+2	AC024592.12	Uncharacterized protein
1+2	NDUFA11	NADH dehydrogenase (ubiquinone) 1 alpha subcomplex
1+2	AC104532.2	Uncharacterized protein
1+2	VMAC	vimentin-type intermediate filament associated coiled-coil protein
1+2	CAPS	calcyphosine

1+2	RANBP3	RAN binding protein 3
1+2	RFX2	regulatory factor X
1+2	ACSBG2	acyl-CoA synthetase bubblegum family member 2
1+2	MLLT1	myeloid/lymphoid or mixed-lineage leukemia (trithorax homolog
1+2	ACER1	alkaline ceramidase 1
1+2	CLPP	ClpP caseinolytic peptidase
1+2	ALKBH7	alkB
1+2	PSPN	persephin
1+2	GTF2F1	general transcription factor IIF
1+2	KHSRP	KH-type splicing regulatory protein
1+2	SLC25A41	solute carrier family 25
1+2	SLC25A23	solute carrier family 25 (mitochondrial carrier
1+2	CRB3	crumbs homolog 3 (Drosophila)
1+2	DENND1C	DENN/MADD domain containing 1C
1+2	TUBB4A	tubulin
1+2	TNFSF9	tumor necrosis factor (ligand) superfamily
1+2	CD70	CD70 molecule
1+2	TNFSF14	tumor necrosis factor (ligand) superfamily
1+2	C3	complement component 3
1+2	GPR108	G protein-coupled receptor 108
1+2	TRIP10	thyroid hormone receptor interactor 10
1+2	SH2D3A	SH2 domain containing 3A
1+2	VAV1	vav 1 guanine nucleotide exchange factor
1	EMR1	egf-like module containing
1	AC025278.1	Uncharacterized protein
1	MBD3L5	methyl-CpG binding domain protein 3-like 5
1	MBD3L4	methyl-CpG binding domain protein 3-like 4
1	MBD3L2	methyl-CpG binding domain protein 3-like 2
1	MBD3L3	methyl-CpG binding domain protein 3-like 3
1	ZNF557	zinc finger protein 557
1	INSR	insulin receptor

Supplementary Table 2. Protein-encoding genes within the deleted interval on 16p13 of patient 2 as indicated by array CGH analysis results annotated using Human Genome Build GRCh37/hg19.

Patient	Gene Name	Description
2	<i>RHBDF1</i>	rhomboid 5 homolog 1 (Drosophila)
2	<i>MPG</i>	N-methylpurine-DNA glycosylase
2	<i>NPRL3</i>	nitrogen permease regulator-like 3 (<i>S. cerevisiae</i>)
2	<i>HBZ</i>	hemoglobin, zeta
2	<i>HBM</i>	hemoglobin, mu
2	<i>HBA2</i>	hemoglobin, alpha 2
2	<i>HBA1</i>	hemoglobin, alpha 1
2	<i>HBQ1</i>	hemoglobin, theta 1
2	<i>LUC7L</i>	LUC7-like (<i>S. cerevisiae</i>)
2	<i>ITFG3</i>	integrin alpha FG-GAP repeat containing 3
2	<i>RGS11</i>	regulator of G-protein signaling 11
2	<i>ARHGDI3</i>	Rho GDP dissociation inhibitor (GDI) gamma
2	<i>PDIA2</i>	protein disulfide isomerase family A, member 2
2	<i>AXIN1</i>	axin 1
2	<i>MRPL28</i>	mitochondrial ribosomal protein L28
2	<i>TMEM8A</i>	transmembrane protein 8A
2	<i>NME4</i>	NME/NM23 nucleoside diphosphate kinase 4
2	<i>DECR2</i>	2,4-dienoyl CoA reductase 2, peroxisomal
2	<i>RAB11FIP3</i>	RAB11 family interacting protein 3 (class II)
2	<i>SOLH</i>	small optic lobes homolog (Drosophila)
2	<i>C16orf11</i>	chromosome 16 open reading frame 11
2	<i>PIGQ</i>	phosphatidylinositol glycan anchor biosynthesis, class Q
2	<i>NHLRC4</i>	NHL repeat containing 4
2	<i>RAB40C</i>	RAB40C, member RAS oncogene family WAP, follistatin/kazal, immunoglobulin, kunitz and netrin domain
2	<i>WFIKKN1</i>	containing 1
2	<i>C16orf13</i>	chromosome 16 open reading frame 13
2	<i>FAM195A</i>	family with sequence similarity 195, member A
2	<i>WDR90</i>	WD repeat domain 90
2	<i>RHOT2</i>	ras homolog family member T2
2	<i>RHBDL1</i>	rhomboid, veinlet-like 1 (Drosophila) STIP1 homology and U-box containing protein 1, E3 ubiquitin protein
2	<i>STUB1</i>	ligase
2	<i>JMJD8</i>	jumonji domain containing 8
2	<i>WDR24</i>	WD repeat domain 24
2	<i>FBXL16</i>	F-box and leucine-rich repeat protein 16
2	<i>METRN</i>	meteorin, glial cell differentiation regulator
2	<i>FAM173A</i>	family with sequence similarity 173, member A
2	<i>CCDC78</i>	coiled-coil domain containing 78
2	<i>HAGHL</i>	hydroxyacylglutathione hydrolase-like
2	<i>NARFL</i>	nuclear prelamin A recognition factor-like

2	<i>MSLN</i>	mesothelin
2	<i>MSLNL</i>	mesothelin-like
2	<i>RPUSD1</i>	RNA pseudouridylate synthase domain containing 1 CTF18, chromosome transmission fidelity factor 18 homolog (S.
2	<i>CHTF18</i>	cerevisiae)
2	<i>GNG13</i>	guanine nucleotide binding protein (G protein), gamma 13
2	<i>PRR25</i>	proline rich 25
