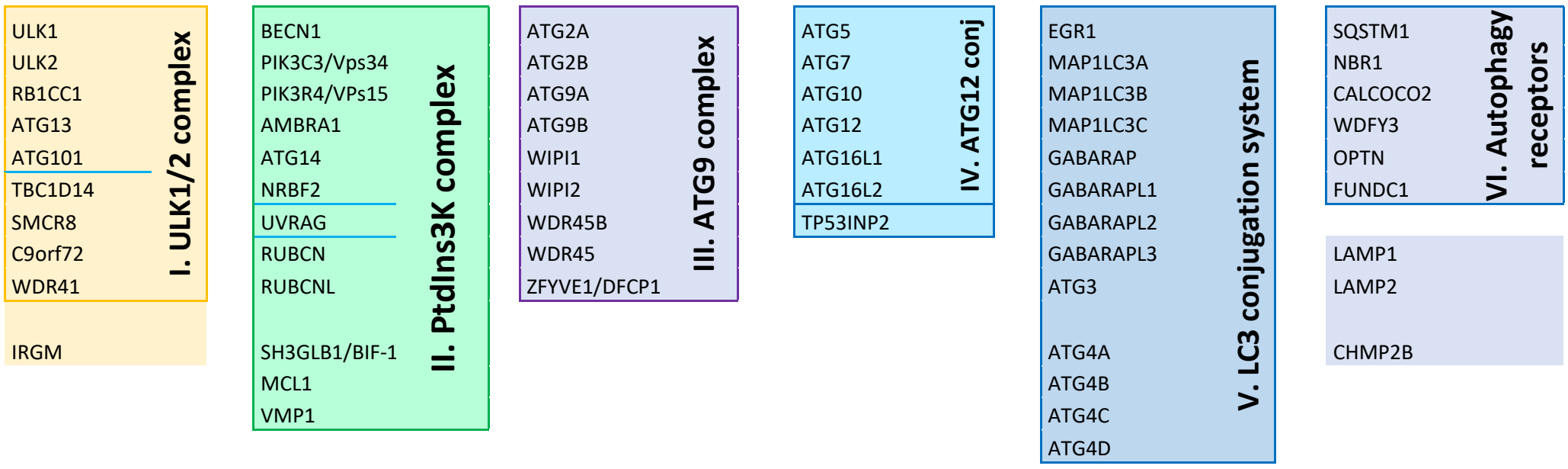
Supplemental Information

An atlas of the genetic variations linking dysregulation of autophagy to human diseases: the missing environmental link

Iris Grosjean and Barnabé Roméo *et al.*

Supplementary list of autophagy-related genes and regulators investigated in this study



Supplementary Figures

Figure S1. Autophagy deficiency in human lung diseases.

Figure S2. Autophagy deficiency in human cardiovascular diseases.

Figure S3. Autophagy deficiency in breast and urogenital diseases.

Figure S4. Autophagy deficiency in human gastrointestinal disorders.

Figure S5. Autophagy deficiency in human central nervous system diseases.

A. Summary of autophagy-related gene variations.

B. Steps of the autophagy pathway affected by SNPs.

C. Phenotypes of autophagy-deficient mouse models.

A

LUNG DISEASES

ASTHMA

ULK1 rs7487166, rs9652059, rs11616018, rs10902472 **RISK**

ATG5 rs573775, rs12201458_ rs12212740 >G rs633724 **RISK**
ATG5 rs510432> A ↑ exp (promoter)
ATG7 rs2594971>GG, rs1375206>CC **RISK**

MAP1LC3B rs8051218, rs7204722 **RISK**

TUBERCULOSIS

ULK1 rs7300908 **RISK**, rs12297124>T ↓ **RISK** **x INFECTION**
ULK1 rs7138581 3'-UTR>C ↓ >CG ↓ **RISK**, ↑ Severity
ULK1 rs9481 3'-UTR >A ↑ **RISK**

IRGM rs4958843>C, rs4958846> C (eQTL prom) ↓ **RISK** ↓ exp
IRGM rs10065172>T (eQTL miR-196) ↓ **RISK** ↓ exp
IRGM rs10059011 ↓ RISK, rs10052068
IRGM rs13361189 , rs9637876
MCL1 rs961581226
ATG4C rs11208029

ATG10 rs1864183, rs3734114

LAMP1 rs9577229

LUNG FIBROSIS

ATG5 rs510432 > T **RISK** **x COAL**
ATG10 rs1864182> GT ↓ **RISK** **x COAL**
ATG12 rs26538>T ↓ exp ↓ **RISK** **x COAL**

COPD

ATG16L1 rs2241880 >A ↑ **RISK** **x SMOKING**
ERG1 rs7729723 >G ↑ **RISK**

NICOTINE DEPENDENCE

GABARAP rs222843 **RISK**
GABARAP rs17710

LUNG CANCER

ULK1 rs7953348>A, rs12303764 >C **THERAGNOSTIC** (platinum)

MCL1 rs3831987 (non-smoker Protective)

BECN1 rs11552193

ATG14 rs17742719>A, rs8003279>G,
rs10099647>A **THERAGNOSTIC** (platinum)

ATG5 rs2245214>C **RISK**
ATG5 rs688810>C **RISK**, BAD **PROGNOSIS** (gefitinib)
ATG5 rs510432 PROTECTIVE, GOOD **PROGNOSIS** (gefitinib)

ATG7 rs8154>T (EGFR*) PROTECTIVE

ATG10 rs10036653>T **RISK** of brain metastasis
ATG10 rs1864183, rs1864182 rs10514231 **RISK**
ATG10 rs1864183 **THERAGNOSIS** (platinum)
ATG10 rs10036653 GOOD **PROGNOSIS** (gefitinib)
ATG10 rs1864182>T GOOD **THERAGNOSIS** (gefitinib)

ATG12 rs26532>AA **RISK** of brain metastasis
ATG12 rs26538>T RISK, BAD **THERAGNOSIS** (gefitinib)
ATG12 rs1058600

ATG16L1 rs2241880 NSCLC >G ↓ RISK of brain metastasis
ATG16L1 rs2241880 (E GFR*) >G **RISK**
ATG16L2 rs11235604 >T BAD **THERAGNOSIS** (gefitinib)
ATG16L2 rs10898880>C **THERAGNOSIS** (radiotherapy) ↑ exp

ATG3 rs13082005>A **THERAGNOSIS** (platinum)
ATG4A rs807185 **RISK**

DRAM rs7955890>A and rs17032060>A **THERAGNOSIS**

B

ULK1/2 complex

ULK1/2 — ASTHMA, TUBERCULOSIS, LUNG CANCER

IRGM — TUBERCULOSIS

LAMP1 — TUBERCULOSIS

PtdIns3K complex-I

BECN1, ATG14, MCL1 — LUNG CANCER, TUBERCULOSIS

ATG12-conj.

ATG5 + ATG12 + ATG16L (1, 2) — ASTHMA, LUNG FIBROSIS, LUNG CANCER; COPD x SMOKING, LUNG CANCER

ATG10 + ATG12 — TUBERCULOSIS, LUNG FIBROSIS, LUNG CANCER

ATG7 + ATG12 — ASTHMA, LUNG CANCER

ATG12 — LUNG CANCER, LUNG FIBROSIS

LC3-conj. system

LC3-II + B + GABARAP — NICOTINE DEPENDENCE

ATG3 — LUNG CANCER

LC3-I

ATG7 — ASTHMA, LUNG CANCER

LC3-I — LUNG CANCER

ATG4A + ATG4C — COPD; EGR1 — TUBERCULOSIS

proLC3B — ASTHMA

C

Ulk1/2$^{-/-}$ (DKO)	**RESPIRATORY distress** [1] Death within 24 h of birth
Atg5$^{-/-}$	
Atg4b$^{-/-}$	**IDIOPATHIC PULMONARY FIBROSIS** (IPF) [2] (tunicamycin) ↑ inflammation
	LUNG FIBROSIS [3] (bleomycin) ↑ inflammation (neutrophil infiltration)
Atg5^{flox} Or *Atg7*^{flox} *Myeloid Cell-Specific*	**FIBROSIS LUNG x INFLAMMATION** [4] ↑ IL18 (bleomycin)
Egr1 KO	**smoke-induced COPD** [5] *Egr1* KO ↓ Atg4B expression ↓ LC3B conversion
Map1lc3b$^{-/-}$	**smoke-induced COPD** [6]
	LUNG FIBROSIS [7] upon aging (bleomycin)
Becn1$^{-/-}$ *Lung-Specific, Conditional*	**LETHAL RESPIRATORY INSUFFICIENCY** [8] ↓ Lung development (branching)
Silencing *Fundc1*	**smoke-induced COPD** [9] cigarette smoke (CSE) COPD mice ↑ FUNDC1. Silencing *Fundc1* → ↓IL6 TNF ↓ CSE-induced mitophagy cell apoptosis. ↑ COPD lung function
Becn1$^{+/-}$	**LUNG ADENOCARCINOMAS** [10,11]
Sh3glb1 $^{-/-}$	**SMALL CELL LUNG CARCINOMAS** [12]

Figure S1_Iris GROSJEAN and Barnabé ROMÉO et al.

A

HEART DISEASES

STROKE

ATG9B rs3800787, rs11769158, rs6464119, rs3918220, rs3763486 (attributed to NOS3)
GABARAPL1 rs3803539

CORONARY ARTERY disease

TBC1D14 rs10804990
ATG9B rs2373929
MAP1LC3 rs2424994, rs6088521

BLOOD PRESSURE and heart rate

ATG9B rs7830 **x AIR POLLUTION** urban **elderly**

Q FEVER x *C. burnetii* infection

ATG5 rs2245214 Protective
MAP1LC3A rs1040747

CARDIOVASCULAR DISEASE

ATG4C rs6683832 _ **ATHEROSCLEROSIS**
ATG4C rs6587988
ATG4D rs10439163
ATG7 rs2594975_ **INFARCTION**
ATG7 rs7635838, rs2447607
ATG9B rs13307588 **INFARCTION**
ATG16L1 rs2241880 RISK ♀
DRAM1 rs77694286_Aging
WDR41 rs335632

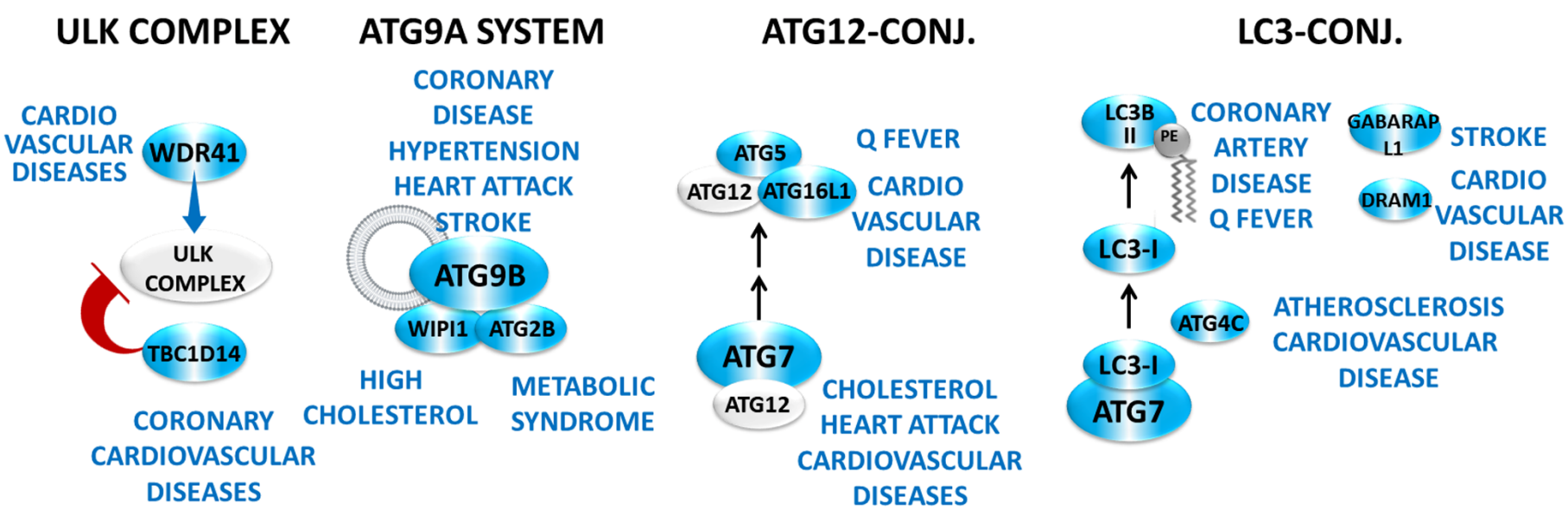
METABOLIC syndrome

ATG2B rs4905480
AMBRA1 rs110389913

HDL cholesterol level

ATG7 rs2606736, rs7635838
WIPI1 rs2909207 eQTL

B



C

Atg13 $^{-/-}$	**HEART defect** [13] embryonic lethality
Rb1cc1 $^{-/-}$	**HEART defect** [14] embryonic lethality
Atg5 $^{Fl/Fl}$ *Cardiac-Specific*	**age-related CARDIOMYOPATHY** [15] ↑ left ventricular dimension ↓ fractional shortening
Uvrag $^{-/-}$	**age-related CARDIOMYOPATHY** [16] ↑ inflammation
Fbxo32/Atrogin1 $^{-/-}$	**CARDIOMYOPATHY** [17] myocardial remodeling, ↓diastolic function, arrhythmias ➔ fails to degrade CHMP2B, resulting in autophagy impairment

Figure S2_Iris GROSJEAN and Barnabé ROMÉO et al.

A

MENOPAUSE

ATG10 rs9447453 C>A **Delayed age**

CANCER

BREAST CANCER

SMCR8 rs1563634 **RISK**

MCL1 rs3831987 **RISK**
NRBF2 rs10995190 **RISK**
NRBF2 rs10509168 **RISK**
VMP1 rs1295925 **RISK**

ATG10 rs1864182 C>A **RISK**
ATG10 rs10514231 C>T **RISK**
ATG10 rs7707921 T>A **RISK**
ATG10 rs73134739 T>C **RISK**

ATG7 rs8154 T>C **PROGNOSIS**

ATG5 rs473543 G>A **THERAGNOSIS**
(Anthracycline and/or taxane)

NBR1 rs17599948 G>C **RISK**

OVARIAN CANCER

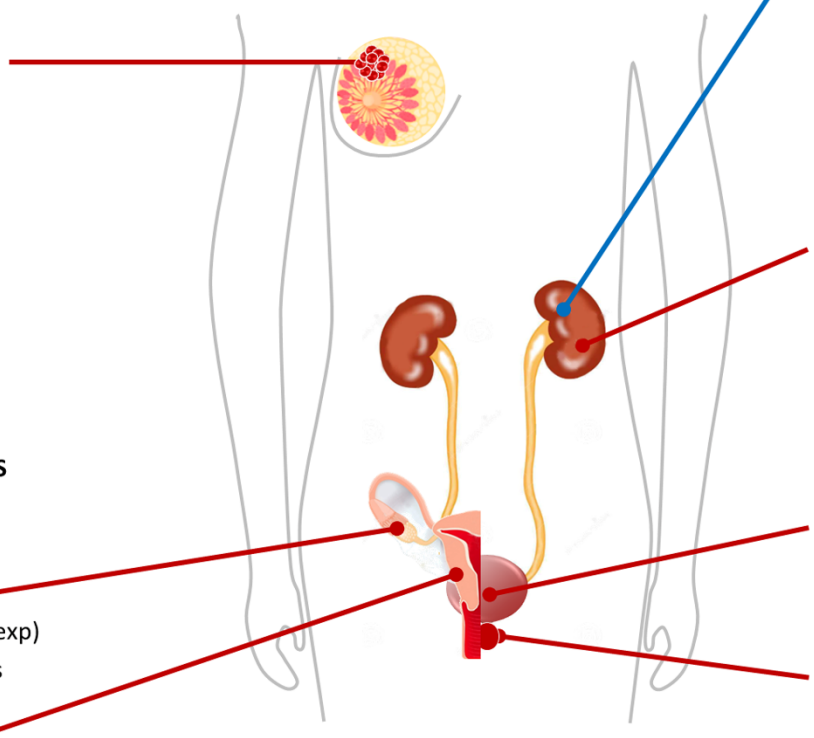
ATG4A rs5973822 (eQTL, miRNA, ↑exp)
SMCR8 rs921986, rs12952556 eQTLs

CERVICAL CANCER

ATG4A rs5973822 (eQTL, miRNA, ↑)
ATG4A rs807181, rs807182 x **HPV INFECTION** RISK
ATG4A rs807183 (eQTL, ↓ exp) x **HPV** RISK

UTERINE LEIOMYOMAS

MAP1LC3C rs1776161 G>A



DISEASES

KIDNEY DISEASE

ATG2A rs188780113 G>A **RISK** of hyperuricemia
Missense rare

ATG16L1 rs2241880 MISSENSE T300A
UROPATHOGENIC E. coli Protection
Protective

ccRCC

ATG4A rs7880351 G>C **THERAGNOSIS** (Pazopanib) ↑ OS PFS
ATG4C rs6683832 G>A rs6670694 G>A
THERAGNOSIS (Pazopanib) ↑ PFS

ATG5 rs490010 A>G **THERAGNOSIS** (Pazopanib) ↓ PFS

ATG16L2 rs10751215 T>C ↓ Metastases

IRGM rs10059011 A>C **RISK** and patient outcome

BLADDER CANCER

ATG2B rs3759601 G>C BAD **THERAGNOSIS** (BCG immunotherapy)

ATG5 rs2245214 C>G BAD **THERAGNOSIS** (BCG immunotherapy)

PROSTATE CANCER

ATG4B rs3771570 C>T

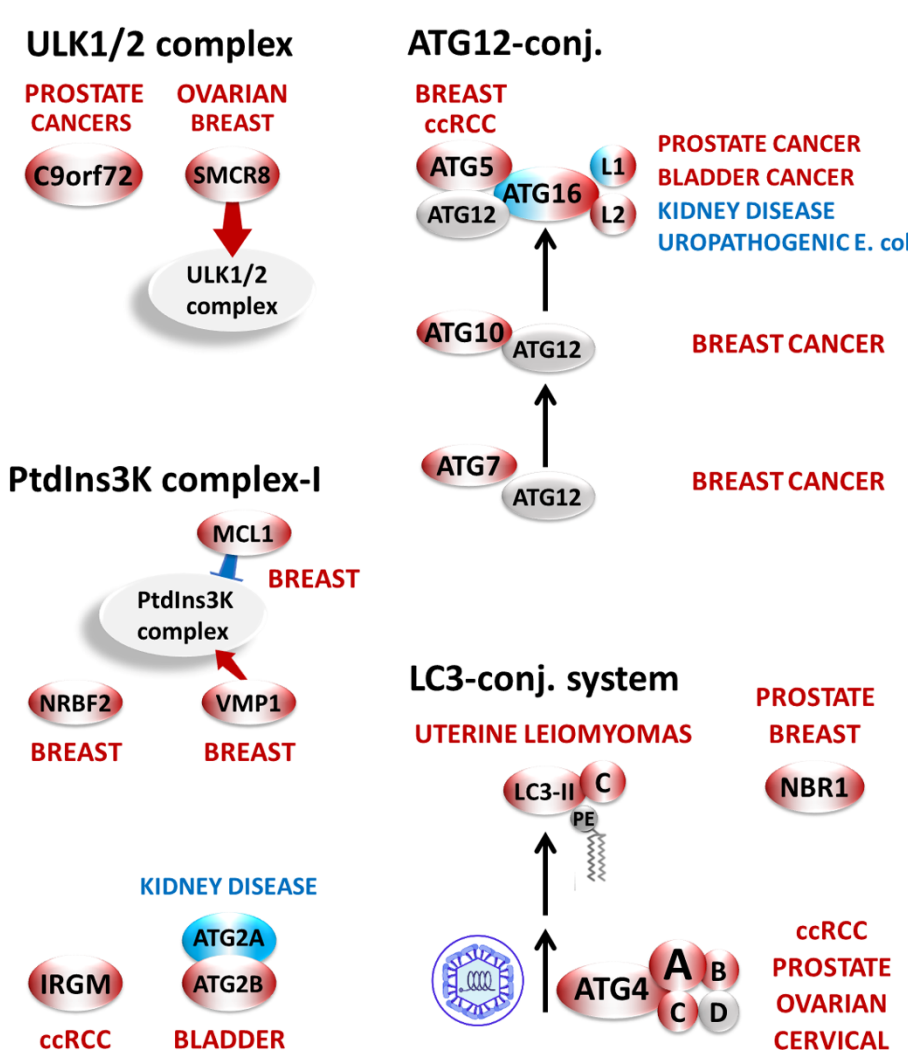
ATG16L1 rs78835907 G>A BAD prognosis

C9orf72 rs774359 T>C, rs3849942 T>C
rs10122902 G>A, rs10757665 T>C

ATG14 rs8015211 T>C aggressivity eQTL

NBR1 rs17599948 G>C BAD **THERAGNOSIS** (radiotherapy)
attributed to BRCA1

B



C

Atg5 $^{-/-}$ *Kidney Specific*	**ACUTE KIDNEY INJURY** [18,19] Cisplatin- and ischemia-reperfusion-induced
Atg7 $^{-/-}$	
Atg5 $^{f/f}$	**KIDNEY DISEASE AND AGING** [20]
Atg16l1	**URINARY TRACT INFECTION** [21]
Atg16l1 T300A	**URINARY TRACT UPEC INFECTION** [21] Protection: ↑ vesicle trafficking, ↓ infection. Independent of autophagy or proinflammatory cytokine responses. ↑ expression of RAB33B, which interacts with ATG16L1, as RAB27B and RAB11A → UPEC exocytosis
Optn $^{-/-}$	***E. Coli*-INDUCED PERITONITIS** [22]
Becn1 $^{+/-}$ Or ***Atg7*** $^{-/-}$ *Ovary Specific*	**REDUCED FERTILITY** [23]
Atg16l1 $^{f/F}$ *Uterus Specific*	**REDUCED FERTILITY**[258] [24] Endometrial Decidualization
Sqstm1 shRNA	**ccRCC growth** [25] addictive to *SQSTM1* gene gain and overexpression *(in vitro* and *in vivo)*[259]
Atg7 $^{f/f}$ ***Pten*** $^{f/f}$ *Inducible Prostate-Specific*	**PROSTATE CANCER** [26] *Atg7* KO delays *Pten*-deficient prostate tumor progression (evidence of ER stress)
Becn1 $^{+/-}$ ***Becn1*** ^{F121A} ***(KI/KI)***	**BREAST CANCER** [10,27-29] ↑ ERBB2/HER2-driven tumorigenesis ERBB2/HER2 interacts with BECN1 and inhibits autophagy Somatic Allelic loss of *BECN1* is observed in over 50% of human breast cancers
Rb1cc1 $^{-/-}$ ***Rb1cc1*** $^{f/+}$ *Breast-Specific*	**BREAST CANCER** [30]
Rb1cc1 $^{f/f}$	**OVARIAN CANCER** [31,32] hyperplasia of epidermis (acanthosis)
Becn1 $^{+/-}$	**EARLY OVARIAN TUMORS** [33]

Figure S3_Iris GROSJEAN and Barnabé ROMÉO et al.

A

INFLAMMATORY DISEASES

TYPE 2 DIABETES

ATG13 rs35619591 G>A, insulin processing
BECN1 rs10512488 G>A
SH3GLB1 rs263436 G>A
DRAM1 rs77694286 A>G

ATROPHIC GASTRITIS

CHMP2B rs1002765 G>A ↑ RISK precancerous

FATTY LIVER DISEASE

IRGM rs13361189 T>C

INFLAMMATORY BOWEL DISEASE

ATG9A rs2382817 A>C

CROHN DISEASE

ULK1 rs3088051 T>C **COMPLICATION,**
ULK1 rs7488085 T>G, rs10902469 G>C Protective
ULK1 rs12303764 T>G, rs11616018 C>T, rs7953348 C>T

IRGM rs4958843, ***r***s4958847 G>A
IRGM rs10065172 C>T - eQTL miR-196 ↑ exp ↑ inflammation
IRGM rs13361189 T>C - eQTL prom - ↓ exp

ATG2A rs17146441 C>T Granulomas

ATG4A rs5973822 A>G - eQTL, miRNA, ↑ exp
ATG4B rs35320439 T>C
ATG4D rs2304165 C>T, rs10439163 A>G – Granulomas

ATG5 rs506027 T>C, rs510432 T>C Theragnosis (anti-TNF) ↑ OS
ATG5 rs9373839 T>C Theragnosis (anti-TNF)

ATG16L1 rs2289476 G>A **x SMOKING**
ATG16L1 rs2241880 A>G **COMPLICATION x SMOKING**
ATG16L1 rs6754677 G>A
ATG16L2 rs11235604 C>T Protective ↓ ATG16L2

CALCOCO2 rs2303015 T>C

CANCERS

HEAD AND NECK CANCERS

MCL1 rs9803935 T>G, rs3738485 C>G,
ATG2B rs3759601 G>C ↑ Pharynx RISK
ATG10 rs1864183 C>T, rs4703533, C>G, rs10514231 C>T
Nasopharynx RISK Bad Theragnosis (Radiotherapy)
ATG12 rs26537 T>C
ATG16L1 rs4663402 A>T, rs2241880 A>G

ESOPHAGEAL CARCINOMA

PI3KC3 rs52911 G>A protective

LIVER CANCERS

ATG5 rs510432 T>C, rs548234 C>T, **HEPATITIS B**
ATG5 rs17067724 A>G, Protective
ATG10 rs1864183 C>T, rs10514231 C>T
ATG12 rs26537 T>C
ATG16L1 rs4663402 A>T, rs2241880 A>G

GASTRIC CANCERS

PI3KC3 rs2162440 A>G, rs7235755 A>G
TELOMERE LENGTH
IRGM rs13361189 T>C Protective x ***H. pylori***
IRGM rs4958847 G>A Protective eQTL ↓ exp
ATG16L1 rs2241880 A>G x ***H. pylori***
CHMP2B rs1002765 G>A x ***H. pylori***

COLORECTAL CANCER

PIK3R4 rs10934954 C>T
ATG16L1 rs2241880 A>G ↓ type I IFN
RB1CC1 rs1129660 A>G **THERAGNOSIS** (anti-VEGF)

B

ULK1/2 complex

ULK1 FIP200 — CROHN, COLORECTAL

IRGM — FATTY LIVER DISEASE, CROHN, GASTRIC CANCER

PtdIns3K complex

T2 DIABETES, CANCERS
BECN1, MCL1, SH3GLB1, PIK3C3, PIK3R4
HNSCC, ESOPHAGEAL, GASTRIC, COLORECTAL

ATG9A system

ATG2 A/B — CROHN, HNSCC
ATG9A — INFLAMMATORY BOWEL DISEASE

ATG12-conj.

CROHN
ATG5, ATG12, ATG16 L1/L2
HNSCC, LIVER
HNSCC, LIVER, GASTRIC, COLORECTAL

DRAM1 — T2 DIABETES
CALCOCO2 — CROHN

ATG10

ATG4 A/B/C/D — CROHN DISEASE

MATURATION

CHMP2B — ATROPHIC GASTRITIS, GASTRIC CANCER

C

Atg7$^{f/f}$ *liver specific*	**STEATOSIS** [34,35] hepatomegaly and benign tumors (adenomas), but not HCCs *Via* SQSTM1/p62-KEAP1-NFE2L2/Nrf2
	HEPATOCELLULAR CARCINOMAS [36] via YY1AP1 autophagy degradation
Sh3glb1/Bif1$^{-/-}$	**HEPATOCELLULAR CARCINOMAS** [12]
Becn1$^{+/-}$	**HEPATOCELLULAR CARCINOMAS** [10,11] Spontaneous and HBV carcinogenesis
Atg16l1$^{f/f}$ *Hematopoietic cells or intestinal epithelium*	**CROHN DISEASE** [37,38] ↑INFLAMMATION ↓Paneth cell granule exocytosis
Atg16l1^{HM}	*Virus + susceptibility gene + commensal bacteria* [39] → ↓ Paneth cell granule secretion UVRAG
	↓Secretory autophagy of lysozyme in Paneth cell [40]
Atg16l1$^{T300A/T300A}$	x **Smoking**, a major CD environmental risk factor [41] ↑Paneth cell apoptosis, metabolic dysregulation, ↓PPARγ pathway (Apoptosis rescued by PPARγ agonist rosiglitazone) CD patients and mice
Irgm2/Lrg47$^{-/-}$	**CROHN DISEASE** [42] ↑ INFLAMMATION ↓Paneth cell granule exocytosis
Optn$^{-/-}$	**bacteria-driven COLITIS** [23] and peritonitis: ↓TNF ↓ Neutrophil recruitment
Atg4b$^{-/-}$	**COLITIS CROHN DISEASE** [43] Paneth cell abnormalities ↑INFLAMMATION
Uvrag^{FS}	**COLORECTAL CANCER** (CRC) [44] Frameshift *UVRAG* mutation → UVRAG^{FS} (truncated) in gastric cancer and CRC; UVRAG^{FS} acts as a dominant-negative mutant
	↑ **age-related TUMORIGENESIS** [45] (CTNNB1/β-catenin stabilization)
	↑ INFLAMMATION in **SEPSIS COLITIS** and colitis-associated cancer development
Sh3glb1/Bif1$^{-/-}$	**DUODENAL ADENOCARCINOMAS** **ESOPHAGEAL SQUAMOUS CELL CARCINOMAS** [12]

Figure S4_Iris GROSJEAN and Barnabé ROMÉO et al.

A

NEURODEGENERATIVE DISEASES

DEMENTIA

C9orf72 rs2282241, rs774359, rs2814707, rs3849942
C9orf72 rs10812615, rs10812616, rs17769294

CHMP2B rs63751048, rs63750652
CHMP2B rs63750653

ALZHEIMER DISEASE

PIK3C3 rs1941526
ATG10 rs4703879, rs891159, rs1485587
WDFY3 rs76117213, rs17009220
DFCP1 rs7155380
RUBCNL rs1408184
SQSTM1 rs104893941

HUNTINGTON DISEASE

ATG7 rs36117895 earlier onset

PARKINSON DISEASE

ULK2 rs281357
ATG7 rs1375206
LAMP1 rs12871648

SPINOCEREBELLAR ATAXIA TYPE 3

BECN1 rs60221525 C>A ↓ BECN1, earlier onset. more severe disease.
BECN1 rs116943570 A>C ↑ BECN1. Neuroprotective effects.

SUPRANUCLEAR PALSY

TBC1D14 rs4689558

NEUROMUSCULAR DISEASES

AMYOTROPHIC LATERAL SCLEROSIS

C9orf72 rs774359, rs2814707, rs3849942, rs3849943
C9orf72 rs2282241, rs3849943, rs10122902, rs17769294

CHMP2B rs63751126

RUBCNL rs2478046

MULTIPLE SCLEROSIS

UVRAG rs80191572 A>G, Theragnosis
EGR1 rs4902647

CANCER

GLIOMA

IRGM rs13361189 >CC ↓ exp → ↑ IFNγ
CALCOCO2 rs8074034

NEUROLOGICAL DISORDERS

SCHIZOPHRENIA

ULK1 rs145451295, rs55815560, rs145279005, rs188342389
PIK3C3 rs1046515390 bipolar disorder
AMBRA1 rs11819869, rs12574668, rs7130141, rs7112229
EGR1 rs11743810
C9orf72 rs10967991

NICOTINE DEPENDENCE

GABARAP rs222843, rs17710

NEURODEVELOPMENTAL DISEASES

NEURAL TUBE DEVELOPMENT

SMCR8 rs12939757
WIPI1 rs77156594, rs146357218

CORTICAL DYSPLASIA AND ATROPHY

PIK3R4 A>C (chr3: 130,681,528) intellectual impairment, ataxia, psychomotor delay, muscle wasting, and late-onset epilepsy

CAUDATE VOLUME

WDR41 rs163030, rs163035, rs335636

MEGALENCEPHALIC LEUKOENCEPHALOPATHY

MCL1 rs1258188045

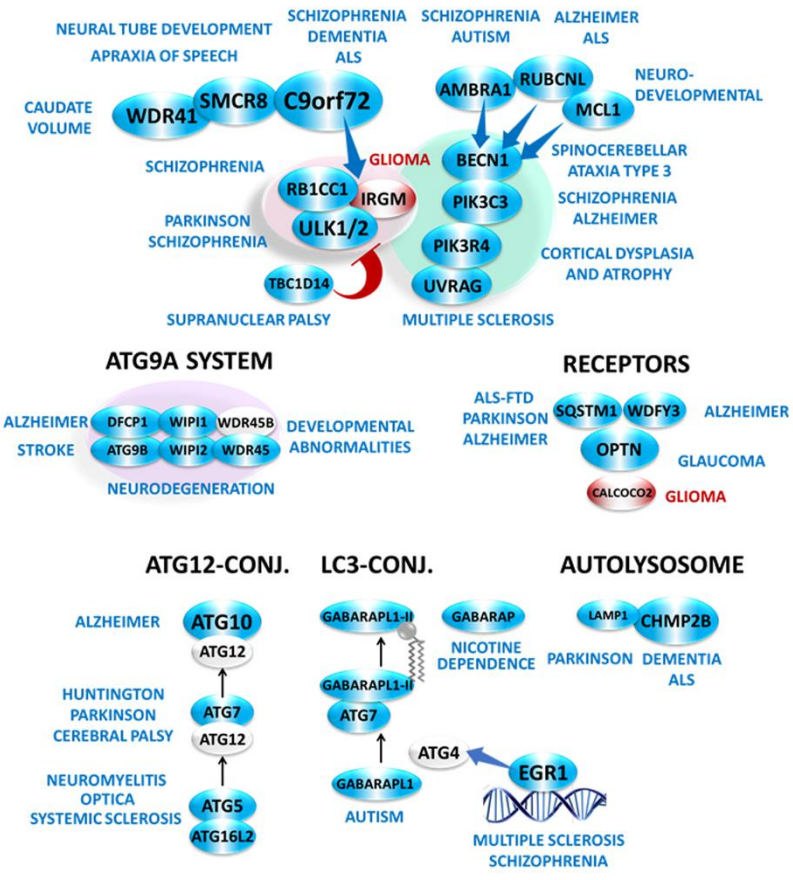
CEREBRAL PALSY

ATG7 rs1470612

AUTISM SYNDROME

AMBRA1 *rs3802890*

B



C

Atg5 $^{-/-}$ ***Atg7*** $^{-/-}$ *Neuron-Specific*	**NEURODEGENERATION** [46,47] deficits in motor function and accumulation of inclusion bodies in neurons
Becn1 $^{+/-}$	**ALZHEIMER DISEASE** [48] ↑ Inflammation IL-1b, IL-18
Sqstm1 $^{-/-}$	**ALZHEIMER-like phenotype** [49] ↑ anxiety, depression, and loss of working memory that precede the accumulation of tau
Chmp2b $^{intron\ 5}$	**FRONTOTEMPORAL** [50,51] C-terminus truncation (Δ Vps4 binding site)
Tsc2 $^{+/-}$	♀ **AUTISM SPECTRUM DISORDERS** [52] hyperactivated mTOR and impaired autophagy
Atg7 **CKO**	
Ambra1 $^{+/-}$	**NEURODEVELOPMENT AUTISM-SPECTRUM DISORDERS** [53] ↓ AMBRA1 protein brain
Wdfy3 $^{+/-}$	**NEURODEVELOPMENTAL delay** [54] (autism/hyperactivity disorder) WDFY3 is required for cerebral cortical size regulation (proper division of neural progenitors) → Wnt-pathway is dysregulated → macrocephaly and deficits in motor coordination and learning.

Figure S5_Iris GROSJEAN and Barnabé ROMÉO et al.

Table S1-S6. Association between common genetic polymorphisms in the autophagy-related genes and human diseases (risk, prognosis, theragnosis). *ATG* SNP (single-nucleotide polymorphism) and their functional annotation (promoter, 5', 3', missense mutations, transcription factor, and miRNA binding sites) were retrieved using PubMed, litvar [58], HaploReg[59,60], and GTEX [61] (eQTLs).

Table S1. **ULK1/2 complex**

Table S2. **PtdIns3K Complex**

Table S3. **ATG9 system**

Table S4. **LC3-conjugation system**

Table S5. **ATG12-conjugation system**

Table S6. **Autophagy receptors**

Related to theragnosis (side effects, toxicity, efficacy), overall survival (OS), and progression-free survival (PFS).

Related to bacterial infection or

viral infection.

Related to air pollution or

coal exposition, or

smoking.

Related to aging

Related to autoimmune and autoinflammatory diseases.

Abbreviations: Alt, alternate allele; Ref, reference allele; exp, expression; (eQTL): eQTL in different tissue of that affected by the disease**;** LD, linkage disequilibrium; MAF, minor allele frequency in European (Eur) or Asian (As) populations; ALS-FTD, amyotrophic lateral sclerosis and frontotemporal degeneration; CD, Crohn disease; ccRCC, clear cell renal cell carcinoma; HCC, hepatocellular carcinoma; HNSCC, head and neck squamous cell carcinoma; NSCLC, non-small cell lung cancer; NPC, nasopharyngeal carcinoma; SLE, systemic lupus erythematosus.

Table S1- *ULK1/2* complex - AUTOPHAGY

Gene	SNP (rs)	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ULK1 **NM_003565.1 93 eQTL**	rs9481	A	G	86%	10	**TUBERCULOSIS** [62] Protective	Asian study	3'UTR	(eQTL)
	rs3088051	T	C	29%	1	**CROHN DISEASE** [63] Complications	Oceanian study	3'UTR	(eQTL)
	rs4964879	G	A	10%	1	**ANKYLOSING SPONDYLITIS** [64] Protective	Asian study	Intronic	eQTL- Nerve
	rs7138581	G	C	13%	5	**TUBERCULOSIS** [62] RISK and SEVERITY	Asian study	3'UTR	(eQTL)
	rs7300908	C	T	2% As 8%	1	**TUBERCULOSIS** [65]	Asians	Intronic	(eQTL) ↑ exp[65]
	rs7487166	A	G	81%	12	**ASTHMA** [66]	American study	Intronic	(eQTL)
	rs7953348	C	T			**CROHN DISEASE** [63] Weak association	Oceanian study		
						NON SMALL CELL LUNG CANCER (NSCLC) [67] Theragnostic (Platinum-based chemotherapy)	Asian study		
	rs9652059	T	C			**ANKYLOSING SPONDYLITIS** [64]	Asian study	Intronic	(eQTL)
						ASTHMA [65]	American study		
	rs11616018	C	T			**ASTHMA** [66]	American study	synonymous	eQTL - colon
						CROHN DISEASE [63] Weak association	Oceanian study		
	rs7488085	T	G	6%	23	**CROHN DISEASE** [63] Protective	Oceanian study	Intronic	(eQTL)
	rs10902469	G	C					5' *ULK1*	(eQTL)
	rs10902472	C	T			**ASTHMA** [66]	American study	Intronic	(eQTL)
	rs12297124	G	T	8%	4	**TUBERCULOSIS** [65] Protective	Asians	Intronic	(eQTL) ↑ exp[65]

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ULK1 **NM_003565.1** **93 eQTL**	**rs12303764**	T	G	36%	1	**CROHN DISEASE** [63,68] **NSCLC** [67] Theragnostic (platinum-based chemotherapy)	European and Oceanian studies Asian study	Intronic	eQTL - colon
	rs55815560	C	T	1%	1	**SCHIZOPHRENIA** [69] A highly heritable psychiatric disorder, combining these 4 SNPs. No variant was individually statistically significant	European study	**MISSENSE** S665L	N/A
	rs145279005	C	T	1%	1			**MISSENSE** A705V	N/A
	rs145451295	C	T	1%	2			**MISSENSE** T242I	N/A
	rs188342389	C	T	0%	6			**Intron** ***MMP17 missense***	N/A
ULK2 **NM_014683** **1450 eQTL**	**rs281357**	T	C	64%	1	**PARKINSON DISEASE** [70]	American study	Intronic	eQTL-Nerve
	rs281366	C	T	3%	3	**ACUTE LYMPHOBLASTIC LEUKEMIA** [71] Theragnostic (asparaginase-associated pancreatitis)	European study	5' *ULK2*	(eQTL)
TBC1D14 **NM_020773** **746 eQTL**	**rs10804990**	G	A	62%	1	**CORONARY HEART DISEASE** [72]	American study	Intronic	eQTL- Artery
ATG13 **NM_014741** **4655 eQTL**	**rs7484002**	A	G	17%	130	**↑ DNA damage** [73]	Asian study	Intronic	eQTL- Skin **↓** exp[73]
	rs35619591	G	A	1%	1	**TYPE 2 DIABETES** [74] Insulin processing	European American study	**MISSENSE** G433R	N/A

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
RB1CC1/FIP200 **NM_014781 68 eQTL**	rs1129660	A	G	21%	19	**COLORECTAL CANCER** [75] Bad theragnosis anti-VEGF-toxicity (bevacizumab)	European study	synonymous	N/A
SMCR8 **NM_144775 1,354 eQTL**	rs1563634	T	C	67%	12	**BREAST CANCERS** [76]	European study	5' *SMCR8*	eQTL-Breast
	rs8080966	C	T	31%	81	**CHILDHOOD APRAXIA OF SPEECH** [77]	American study	**MISSENSE** P524L	eQTL- Brain
	rs12939757	A	G			**NEURAL TUBE DEFECTS** [78] Low maternal folate intake	American study	3' *SMCR8*	eQTL- Brain
	rs921986	C	T			**OVARIAN CANCER** [79]	American study	3' *SMCR8*	eQTL- ovary
	rs12952556	T	C					3' *SMCR8*	eQTL- ovary
WDR41 **NM_018268 10007 eQTL** **ULK1 partner**	rs163016	A	T	40%	41	**MYOPIA** [80]	World collaboration	Intronic	(eQTL)
	rs163030	A	C	48%	83	**CAUDATE VOLUME** [81] Brain region implicated in common neurological and psychiatric disorders	American study	Intronic	eQTL - Brain
	rs163035	A	G					Intronic	eQTL - Brain
	rs335636	A	G					Intronic	eQTL - Brain
	rs335632	C	T	1%	1	**HEART DISEASES** [82]	World collaboration	N/A	(eQTL)
	rs33204	C	T	40%	28	**MYOPIA** [80]	World collaboration	**MISSENSE** V326I	(eQTL)
	rs10514104	T	C	18%	14	Age-related **HEARING IMPAIRMENT** [83]	European study	Intronic	eQTL - Brain ↓ exp [83]

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
C9orf72 **NC_000009.12** **6048 eQTL** **ULK1 partner**	**rs774359**	T	C	27%	81	**ALS** **FTD** [84–86] **PROSTATE CANCER** [87]	Asian and American Word collaboration American study	3'UTR	eQTL - Brain
	rs2814707	C	T			**ALS-FTD** [84–86] **SYSTEMIC LUPUS ERYTHEMATOSUS (SLE)**[88] high serum IFNK (type I IFN)	Asian and American World collaboration American study	5' *C9orf72*	eQTL - Brain
	rs3849942	T	C			**ALS-FTD** [89] **PROSTATE CANCER** [87]	European study American study	3' *C9orf72*	eQTL - Brain
	rs3849943	C	T	75%	1	**ALS** [90]	European study	3' *C9orf72*	eQTL - Brain
	rs2282241	C	A	43%	6	**ALS-FTD** [89]	European study	Intronic	eQTL - Brain
	rs2492816	G	A	42%	8	**SLE** [91]	European study	Intronic	(eQTL)
	rs3849944	T	C	47%	10	**SLE**[88] high serum IFNκ	American study	3' UTR	(eQTL)
	rs10812615	T	C			**FTD** [92]	Oceanian study	Intronic	eQTL- Brain
	rs10812616	T	A					Intronic	eQTL - Brain
	rs10122902	G	A	16%	9	**ALS** [93] **PROSTATE CANCER** [87]	American study American study	synonymous	(eQTL)
	rs10757665	T	C	25%	25	**PROSTATE CANCER** [87]	American study	Intronic	(eQTL)
	rs10967991	C	T			**SCHIZOPHRENIA** [94]	Asian study	5' *C9orf72*	(eQTL)
	rs12686452	T	C	21%	7	**SLE**[88] ♀ high serum IFNK	American study	Intronic	(eQTL)
	rs17769294	T	C	11%	10	**ALS** [93] **FTD** [92]	Oceanian study American study	**MISSENSE** N207S	(eQTL)

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
IRGM **NM_001145805 1645 eQTL**	rs4958843	T	C	9%	168	**TUBERCULOSIS & CROHN DISEASE** [95] ↓ RISK	Middle East study	5' *IRGM*	eQTL – Lung ↑ exp[95]
	rs10065172	C	T			**CROHN DISEASE** [96–100] ↓ IRGM mRNA in patient blood and ileum[39], ↑ inflammation	American studies	Synonymous ***MIR196* binding seed** ↑ IRGM [101]	eQTL Lung Colon Blood ↑ exp [96] ↓ exp [97,100,102] = exp [103]
						GRAVE DISEASE [104]	Asian study		
						SEPSIS [102] ↑ mortality	Asian study		
						TUBERCULOSIS [103–106] ↓ RISK in Asians, but not in Africans/African-Americans	Asian study		
	rs11747270	A	G			Severe **PERIODONTITIS** [107]	European study	N/A	(eQTL)
	rs13361189	T	C			**CROHN DISEASE** [96,97,100,108] ↑ inflammation CD colitis (lactoferrin in ileum and TNF in blood) (GTEX) [39]	American study European study	5' *IRGM*	**PROMOTER** eQTL - Lung Colon ↓ mRNA in blood and ileum [97,100]
						GASTRIC CANCER [109] ↓ RISK x ***H. pylori***	Asians Ocean study		
						GLIOMA [110] ↑ IFNG ↑ serum IL4	Asian study		
						GRAVE DISEASE [104]	Asian study		
						LEPROSY [111] ↑ ***Mycobacterium leprae*** ↑ INFG and IL4	Asian study		
						PERIODONTITIS [107]	European study		
						NON-ALCOHOLIC FATTY LIVER DISEASE [112,113]	Asian study American study		
						TUBERCULOSIS [105]	Asian study		

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
IRGM **NM_001145805 1645 eQTL**	rs4958846	T	C	13%	51	**TUBERCULOSIS** [95,106,114] ↓ RISK	Middle East and Asian studies	5' *IRGM*	eQTL - Lung ↓ exp [34]
	rs4958847	G	A			**CROHN DISEASE** [108]	European study	Intronic	eQTL - colon
						GASTRIC CANCER [109,115] ↓ RISK (intestinal type)	Asian and European studies		
						GRAVE DISEASE [104]	Asian study		
						PERIODONTITIS [107]	European study		
	rs9637876	C	T	9%	163	**TUBERCULOSIS** [116]	African and European study	5'UTR	eQTL - lung ↑ exp[116]
	rs10052068	T	C	0%	1			Intronic	N/A
	rs10059011	A	C	54%	1	**METASTATIC CLEAR CELL RENAL CELL CARCINOMA ccRCC** [117] RISK and patient outcome	European study	5'UTR	(eQTL)
						TUBERCULOSIS [115]	African and European study		

Table S2 – PtdIns3K Complex AUTOPHAGY_LAP

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
PIK3C3 **Vps34** **NM_002647** **3755 eQTL**	rs52911	G	A	34%	47	**ESOPHAGEAL SQUAMOUS CELL CARCINOMA** [118] Protective	Asian study	Intronic	eQTL - Esophagus
	rs1941526	C	T	20%	83	**ALZHEIMER DISEASE** [119]	American study	Intronic	eQTL - Nerve
	rs2162440	A	G	79%	25	**GASTRIC CANCER** [120,121] (GASTRIC CARDIA ADENOCARCINOMA) associated with shorter telomeres	World collaboration Asian study	3' *PIK3C3* (5' MIR4318)	N/A
	rs7235755	A	G						N/A
PIK3R4 **Vps15** **NM_014602** **263 eQTL**	rs10934954	C	T	20%	176	**COLORECTAL CANCER** [122]	European study	Intronic	eQTL - Colon
	rs2200368	A	G			**AGE-RELATED MACULAR DEGENERATION** [123]	American study	Intronic	(eQTL)
	rs11713445	G	A					Intronic	(eQTL)
BECN1 **NM_003766** **157 eQTL**	rs10512488	G	A	21%	1	**TYPE 2 DIABETES** [124] **NON-HODGKIN LYMPHOMA** [125]	World collaboration American study	Intronic	eQTL - Blood
	rs11552193	C	T	0%	1	**NSCLC** [126]	American and European study	3'UTR	Not found miRNA Target Sites
	rs60221525	C	A	6%	1	**MACHADO-JOSEPH DISEASE/ SPINOCEREBELLAR ATAXIA TYPE 3** [127] Neuroprotective	European study	3' *BECN1*	Not found ↑ exp[127]
	rs116943570	A	C	0%	1			3' *BECN1*	Not found
NRBF2 **NM_030759** **1679 eQTL**	rs10995190	G	A	14%	11	**MAMMOGRAPHIC DENSITY** [128] Putative Enhancer at the 10q21.2 **BREAST CANCER** Risk Locus Regulate NRBF2 Expression	European study	ZNF365 Intronic	N/A
	rs10509168	T	C	52%	4			ZNF365 Intronic	N/A

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
UVRAG **NM_003369 1790 eQTL Autophagosome maturation**	rs80191572	A	G	5%	1	**MULTIPLE SCLEROSIS** [129] Theragnosis (Copaxone)	World collaboration	Intronic	(eQTL)
	rs7933235	A	G	8%	97	**VITILIGO** [130] LD with rs7118567 MISSENSE P10H	Asian study	Intronic	Not found
	rs1458836	C	T					5' *UVRAG*	eQTL - Skin
	rs7116263	C	G	8%	9	**CANCER : Etoposide-induced cytotoxicity** [131]	American study	Intronic	(eQTL)
	rs17134573	G	A	3%	154	**Blood lipid, body mass index** [132]	American study	Intronic	(eQTL)
	rs594826	G	A	6%	3			Intronic	N/A
RUBCNL **NM_025113 2581 eQTL**	rs1408184	C	T	34%	72	**ALZHEIMER DISEASE** [133]	European study	**MISSENSE** G152R	eQTL - Brain
	rs2478046	G	A	14%	61	**ALS** [134]	European study	3' *RUBCNL*	eQTL - Nerve
MCL1 **NM_008562.3 402 eQTL Inhibitor of BECN1**	rs1258188045	C	G, T	0%	1	**MEGALENCEPHALIC LEUKOENCEPHALOPATHY** [135]	European study	**MISSENSE** R84C	Not found
	rs9803935	T	G	56%	36	**HNSCC** [136] **xHPV16-related** oro-pharyngeal type	American study	5' *MCL1*	(eQTL)
	rs3738485	C	G					5' *MCL1*	(eQTL)
	rs961581226	C	A	0%	1	**TUBERCULOSIS** [137]	Asian study	5' *MCL1*	Not found
	rs3831987	CC	21mer	N/A	1	**LUNG CANCER in non-smoker** [138] Protective **BREAST CANCER** [139] Protective	Asian study Asian study	5' *MCL1*	Not found ↑ exp
VMP1 **NM_030938 272 eQTL Partner of PI3K**	rs1295925	T	C	71%	32	**BREAST CANCER** [140] Protective **OSTEOSARCOMA** [141] Protective - TP53 transcriptional binding site	Asian study Asian study	Intronic	eQTL - Breast

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ATG14 **NM_014924 8192 eQTL**	rs8015211	T	C	35%	53	**PROSTATE CANCER** [142] aggressivity (GWAS) not validated in an independent study	American study	intronic	eQTL Prostate
	rs8003279	A	G			**NSCLC** [67] good Theragnosis (platinum)	Asian study	synonymous	(eQTL)
	rs1009647	G	A	27%	9			3' of TBPL2	(eQTL)
	rs17742719	G	T	18%	8			intronic	eQTL Lung
SH3GLB1/ BIF-1 **NM_016009** ***849 eQTL***	rs263436	G	A	24%	35	**TYPE 2 DIABETES** [143] GWAS	American studies	intronic	N/A
AMBRA1 **NM_017749.3 148 eQTL**	rs11038913	T	C	8%	1	**ARTERY DISEASE** [144] associated with blood proinsulin levels	European study	Intronic	eQTL - Artery
	rs3802890	A	G	31%	1	♀ **AUTISM**[145] protective GWAS	European Study(US and European GWAS)	Intronic ↓ mRNA	(eQTL)
	rs7130141	C	T	17%	113	**SCHIZOPHRENIA**[146]	European Study	Intronic	eQTL - Brain
	rs7112229	C	T					Intronic	eQTL - Brain
	rs11819869	C	T					Intronic	eQTL - Brain

III - ATG9 system AUTOPHAGY

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ATG2A **NM_015104 16 eQTL**	rs17146441	C	T	23%	2	**CROHN DISEASE** [147] Granulomas	European study	Intronic	(eQTL)
	rs188780113	G	A	0%	1	**KIDNEY DISEASE** [148] RISK of hyperuricemia	Asian study	**MISSENSE** R478C	N/A
ATG2B **NM_018036 318 eQTL**	rs3759601	G	C	42%	27	**BLADDER CANCER** [149] Theragnosis (BCG immunotherapy) ↓ autophagosome formation (macrophages) **HNSCC**[150] ↑ RISK pharyngeal cancer	European study European study	**MISSENSE** Q1383E	(eQTL)
ATG9A **NM_024085 147 eQTL**	rs2382817	A	C	64%	74	**INFLAMMATORY BOWEL DISEASE** [151]	European study	PNKD intron	eQTL – Colon
ATG9B **NM_173681 283 eQTL**  **Attributed to NOS3**	rs7830	G	T	36%	1	**BLOOD PRESSURE** [152] **x AIR POLLUTION** - urban elderly	Asian study	5' *ATG9B*	eQTL - Artery
	rs2373929	G	A	46%	1	**CORONARY ARTERY DISEASE** [153]	Middle East study	Intronic	eQTL - Artery
	rs3763486	T	C	20%	2	**SKIN BASAL CELL CARCINOMA** [154] **STROKE** [155]	American studies	5' *ATG9B*	(eQTL)
	rs3800787	G	C	40%	1	**STROKE** [155]		Intronic	N/A
	rs3918220	C	G	0%	1			Intronic	N/A
	rs6464119	T	C	77%	2			Intronic	eQTL - Artery
	rs11769158	A	G	88%	1			5' *ATG9B*	(eQTL)
	rs11760487	G	A	13%	2	**BONE DENSITY** [156]	American study	Intronic	(eQTL)
	rs12666075	G	T	19%	4			Intronic	Not found
	rs13307588	A	G	94%	2	**MYOCARDIAL INFARCTION** [157]	European study	Intronic	(eQTL)

Gene	variant	Ref	Alt	EUR freq	LD	Disease Susceptibility	Populations	Funct Annot	eQTL GTEX
WIPI1 **NM_017983 1114 eQTL**	rs2909207	T	C	76%	27	**Age-adjusted medium HDL particles** [158]	American and European studies	Intronic	(eQTL)
	rs77156594	A	G	0%	1	**ANENCEPHALY** [159]	American study	**MISSENSE** L406P	Not found
	rs146357218	C	T	0%	1			**MISSENSE** R328Q	Not found
WIPI2 **NM_015610**	**DEVELOPMENTAL ABNORMALITIES** [160]mental retardation, neurological, psychiatric, skeletal and cardiac abnormalities. homozygous (c.G745A ; pV249M). ↓ Binding of the V231M mutant to ATG16L1 (ATG5-12), ↓ WIPI2 puncta, ↓ LC3 lipidation and ↓ autophagic flux.								
WDR45 **NM_007075.3**	♀ **Neurodegeneration with BRAIN IRON ACCUMULATION** [161] **BETA-PROPELLER PROTEIN-ASSOCIATED NEURODEGENERATION**[162] WDR45 mutations →↓ autophagy degradation of ferritin→↑ **iron in the brain** (basal ganglia) X-linked								
ZFYVE1/DFCP1 **NM_021260 1079 eQTL**	rs7155380	T	C	45%	25	**ALZHEIMER DISEASE** [163] Late-Onset	American study	Intronic	eQTL - Brain/Nerve
	ZFYVE1/DFCP1 (WIPI2 partner) associates with lipid droplets.[164]								

IV LC3-conjugation system LAP AUTOPHAGY

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ATG3 **NM_022488 98 eQTL**	rs13082005	G	A	40%	4	**NSCLC** [67] Theragnosis (Cisplatin)	Asian study	Intronic	(eQTL)
ATG4A **NM_178270 309 eQTL**	rs807181	G	C	45%	2	**CERVICAL CANCER** [165] **↑ HPV INFECTION** RISK - **↓** autophagy	Asian study	Intronic	(eQTL)
	rs807183	G	A					Intronic	(eQTL)
	rs807185	A	T	64%	1	**LUNG CANCER** [166]	Asian study	Intronic	(eQTL)
	rs5973822	A	G	6%	1	**CERVICAL CANCER** [165] **CROHN DISEASE** [147] Granulomas **OVARIAN CANCER** [167]	Asian study European study Asian study	3'UTR miRNA binding site	(eQTL)
	rs7880351	G	C	45%	1	**ccRCC** [117] Theragnosis (Pazopanib) **↑** OS PFS	European study	Intronic	(eQTL)
ATG4B **NM_013325.5 1758 eQTL**	rs3771570	C	T	13%	21	**PROSTATE CANCER** [168]	World collaboration	5' *ATG4B*	(eQTL)
	rs35320439	T	C	33%	1	**CROHN DISEASE** [169]	European and Asian study	5' *ATG4B*	(eQTL)
ATG4C **NM_032852 2531 eQTL**	rs11208029	A	G	14%	109	**TUBERCULOSIS** [170]	African study	Intronic	(eQTL)
	rs6587988	C	T			**CARDIOVASCULAR DISEASES** [144] total cholesterol, triglycerides	European study	Intronic	eQTL - Artery **↓** exp [171]
	rs11208030	G	A			**KASHIN-BECK DISEASE** [171]	Asian study	Intronic	eQTL – Heart **↓** mRNA / protein exp[171]
	rs4409690	G	A			**KASHIN-BECK DISEASE** [171]	Asian study	Intronic	(eQTL) **↓** exp[171]
	rs12097658	T	C					Intronic	

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ATG4C **NM_032852 2531 eQTL**	rs6670694	G	A	42%	96	**ccRCC** [117] Theragnosis (Pazopanib) ↑ PFS	European study	Intronic	(eQTL)
	rs6683832	G	A	56%	33	**ATHEROSCLEROSIS** [172] **ccRCC** [117] Theragnosis (Pazopanib) ↑PFS	American study European study	Intronic	eQTL - Artery
ATG4D **NM_032885 274 eQTL**	rs2304165	C	T	15%	16	**CROHN DISEASE** [147] Granulomas	European study	synonymous	(eQTL)
	rs7248026	T	G	25%	31			5' *ATG4D*	(eQTL)
	rs10439163	A	G	40%	23	**CARDIOVASCULAR DISEASES** [144] LDL and cholesterol **CROHN DISEASE** [147] Granulomas	European study European study	5' *ATG4D*	eQTL - Heart, Colon
EGR1 **NM_001964.2 8 eQTL**	rs4902647	C	T	46%	22	**MULTIPLE SCLEROSIS** [173]	American study	3' *EGR1*	(eQTL)
	rs7729723	A	G	36%	9	**CHRONIC OBSTRUCTIVE PULMONARY DISEASE (COPD)** [5,174] **X Smoking**-induced disease [105]	Asian study American study	5' *EGR1*	(eQTL)
	rs11743810	C	T	54%	7	**SCHIZOPHRENIA** [175]	Asian study	Intronic	(eQTL)
GABARAP **NM_007278.1 37 eQTL**	rs222843	T	C	38%	22	**NICOTINE DEPENDENCE** [176] **x Smoking**-induced disease **NICOTINE AND ALCOHOL DEPENDENCE** [177]	American study Asian and American study	**PROMOTER** (MISSENSE CLDN7)	eQTL - Brain/Nerve ↑ exp [177]
	rs17710	A	T	15%	2	**NICOTINE DEPENDENCE** [176,177]	American	**3'-UTR**	(eQTL)
GABARAPL2 **NM_007285 97 eQTL**	rs12599322	G	A	5%	26	**DNA DAMAGE** [73]	Asian study	RP11-77K12.8	eQTL
GABARAPL3 **NM_028287 15 eQTL**	rs6496667	C	A	20%	110	**RHEUMATOID ARTHRITIS** [178]	Asian study	5' *GABARAPL3*	(eQTL)

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
GABARAPL1 ***NM_031412*** ***257 eQTL***	in **AUTISM SPECTRUM** disorder-associated pathways. deletions of GABARAPL1 in autism [179] GABARAPL1 is the **most highly expressed gene** among the family in the **central nervous system**.								
MAP1LC3A **NM_181509** **27368 eQTL**	rs1040747	C	G	36%	43	**CHRONIC Q FEVER** [180] ***x C. burnetii***-induced cytokine production - Protective	European study	3'UTR	eQTL - Blood
	rs2424994	C	T	15%	1	**CORONARY ARTERY DISEASE** [144]	European study	5' *MAP1LC3A*	eQTL - Heart
	rs6088521	A	C	49%	1			5' *MAP1LC3A*	eQTL - Heart
MAP1LC3B **NM_022818** **1437 eQTL**	rs8051218	T	C	6%	4	**ASTHMA** [66]	American study	Intronic	(eQTL)
	rs7204722	C	T	82%	2			Intronic	(eQTL)
	rs933717	T	C	44%	26	**SLE**[181]	Asian study	3' *MAP1LC3B*	eQTL – Blood ↑ exp[181]
MAP1LC3C **NM_001004343** **2 eQTL**	rs1776161	G	A	42%	1	**UTERINE LEIOMYOMAS** [182] Clinical Trial	American study	Exon1	N/A

Several ***cancer-associated mutations in LC3*** have been reported that either attenuate its binding to **ATG7** and subsequent lipidation (*LC3B* **Y113C**) [183] or its interaction and cleavage by **ATG4** (*LC3A* **R70C/H** rs778324131, *LC3B* **T113** rs200708875, and *LC3C* **R76C** rs776473823) [184]. These ***loss-of-function LC3 mutations*** could confer tumor-promoting features as they are reducing but not completely blocking autophagy in cells, and thus still support some level of autophagy for the continuous growth of tumor cells at later stages. Melanoma/hepatocellular carcinoma [185,186].

GABARAPs and LC3s have opposite roles in regulating ULK1 for autophagy induction [187]

It has been reported that mutation of the ULK1 LIR that disrupts the Atg8-family protein-ULK1 interaction drastically reduces the activity of ULK1, autophagic degradation of SQSTM1, and phagophore formation in response to starvation. Similarly, disruption of the ATG13-Atg8-family protein interaction suppresses ULK1 activity and autophagosome formation. By reconstituting Atg8-family protein-depleted cells with individual Atg8-family members, it has been shown that:

- The GABARAP subfamily (GABARAP and GABARAPL1) positively regulates ULK1 activity and autophagosome formation in response to starvation.
- In contrast, the LC3 subfamily (LC3B and LC3C) negatively regulates ULK1 activity and phagophore formation.

V - *ATG12*-conjugation system.

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
	rs490010	A	G	48%	20	**ccRCC** [117] Theragnosis (Pazopanib) ↓PFS	European study	Intronic	(eQTL)
	rs473543	**A**	**G**			**BREAST CANCER** [188] Theragnosis (Anthracycline and/or taxane) ↑PFS	Asian study	5' *ATG5*	(eQTL)
	rs506027	**T**	**C**			**CROHN DISEASE** [189] Theragnosis (anti-TNF) ↑ OS **SEPSIS** [190] ↑ TNF IL1B	European study Asian study	5' *ATG5* promoter	(eQTL) ↓ exp [190] ↑ exp [191–193]
						ASTHMA [190–192] ↑ neutrophils	American and Asian studies		
ATG5 **NM_004849.2** **509 eQTL**	rs510432	T	C	44%	12	**APLASTIC ANEMIA** [194] ↓ RISK	Asian study	5' *ATG5* promoter	(eQTL) ↓ exp [190] ↑ exp[124] ↑ exp[191–193]
						ASTHMA [191–193] ↑ neutrophils	American and Asian studies		
						CROHN DISEASE [189] Theragnosis (anti-TNF) ↑ OS	European study		
						HCC [195] **HBV-related diseases**	Asian study		
						NSCLC EGFR* [196] Theragnosis (gefitinib) ↑ OS PFS	Asian study		
						LUNG FIBROSIS [197] ↓ RISK x **COAL**	Asian study		
						MELANOMA [198] Bad prognosis ↓ tumor-infiltrating lymphocytes	American study		
						SEPSIS [190] ↑ TNF IL1B	Asian study		

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ATG5 **NM_004849.2 509 eQTL**	rs2245214	C	G	36%	11	**BLADDER CANCER** [199] autophagy (macrophages) ↓ BCG vaccine-induced trained immunity	Middle East study	Intronic	(eQTL)
						MELANOMA [198] Bad prognosis ↓ tumor-infiltrating lymphocytes	American study		
						NSCLC [149]	European study		
						PAGET DISEASE [200]	European study		
						Q FEVER [180] **x *C. burnetii* infection** - Protective	European study		
						SLE[201,202]	American studies		
						SYSTEMIC SCLEROSIS [203]	World		
						THYROID CANCER [204]	European study		
	rs633724	C	T			**ASTHMA** [66]	American study	Intronic	(eQTL)
	rs573775	G	A	28%	1	**ASTHMA** [193]	Asian study	Intronic	(eQTL) ↑ exp (PBMC) [205]
						APLASTIC ANEMIA [194]	Asian study		
						BEHÇET DISEASE [205] ↓ RISK	Asian study		
						SLE[202,206,207] ↑ IFNA IL10	American study and European studies		

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ATG5 ***NM_004849.2*** ***509 eQTL***	rs548234	C	T	68%	5	**HCC** [208] Associated with Chronic Hepatitis B virus infection **HEPATITIS B** virus infection [208] **NEUROMYELITIS OPTICA** [209] RISK of demyelination ↑T cell exp **SLE** [203,210] Protective	Asian study Asian study Asian study and World collaboration	3' *ATG5*	N/A
	rs688810	A	G	22%	1	**NSCLC EGFR*** [196] Bad theragnosis (Gefitinib) ↓PFS	Asian study	5' *ATG5*	(eQTL)
	rs803360	G	C	48%	26	**ACQUIRED APLASTIC ANEMIA** [194] Protective	Asian study	Intronic	(eQTL)
	rs3827644	G	C			**SYSTEMIC SCLEROSIS** [203,211]	European study World	Intronic	(eQTL)
	rs9372120	T	G	19%	31	**MULTIPLE MYELOMA** [212] **RHEUMATOID ARTHRITIS** [213] **SYSTEMIC SCLEROSIS** [203]	American study American study World collaboration	Intronic	(eQTL)
	rs9373839	T	C			**CROHN DISEASE** [189] Theragnosis (anti-TNF) **SYSTEMIC SCLEROSIS** [203]	American study World	Intronic	(eQTL)
	rs6568431	A	C	60%	14	**CHRONIC HEPATITIS B VIRUS (HBV) INFECTION** [208] **SLE** [201,202] ↑ anemia and renal involvement (PBMC)	American studies	3' *ATG5*	N/A ↑ exp[208]
	rs6937876	G	A			**AUTOIMMUNE DEMYELINATING DISEASE** [194,209] Protective	Asian studies		N/A
	rs12201458	**C**	**A**	12%	2	**ASTHMA** [191,192]	American studies	Intronic	N/A
	rs12212740	**G**	**A**	13%	4	**ASTHMA** [66,193] Protective	American and Asian studies	Intronic	N/A
	rs17067724	A	G	1%	1	**HCC** [214] Protective	Asian study	Intronic RP1-60O19.1	N/A

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ATG7 **NM_001144912 5828 eQTL**	rs8154	T	C	31%	1	**LUAD (EGFR*)** [196] **BREAST CANCER** [215] Bad prognostic ↓ PFS and OS	Asian study Asian study	Synonymous methylation meQTL[215]	eQTL – Lung ↑ exp[215]
	rs1375206	C	G	60%	36	**ASTHMA** [193] ↓serum IL8 levels **PARKINSON disease** [216]	Asian study Asian study	Intronic	(eQTL)
	rs1470612	C	T	18%	77	**CEREBRAL PALSY** [217]	Asian study	Intronic	(eQTL)
	rs2594971	G	A	65%	45	**ASTHMA** [193] ↓serum IL8 levels	Asian study	5' *ATG7*	(eQTL)
	rs2594975	T	C	51%	1	**MYOCARDIAL INFARCTION** [218] ↑ binding of transcription factors, transcription	Asian study	5' *ATG7*	Not found
	rs7635838	T	C	59%	35	**CARDIOVASCULAR DISEASE** [144] HDL	European study	Intronic	(eQTL)
	rs2447607	C	G,T	59	14	**Systolic BLOOD PRESSURE** [144]	European study	Intronic	eQTL muscle
	rs36117895	T	C	3%	56	**HUNTINGTON DISEASE** [219]	European study	**MISSENSE** V471A	(eQTL)
ATG10 **NM_001131028 16437 eQTL**	rs1864182	C	A	58%	11	**BREAST CANCER** [220] Protective	Asian study	**MISSENSE** P220H	eQTL - Lung
						NSCLC [221] Poor survival	Asian study		
						NSCLC EGFR* [196] Theragnosis (Gefitinib) ↓ RISK for acquired RESISTANCE. ↑ RISK of primary RESISTANCE	Asian study		
						LUNG FIBROSIS [197] Protective **x COAL**	Asian study		
						MELANOMA [198] ↑ tumor-infiltrating lymphocytes (TILs)	American study		

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ATG10 **NM_001131028 16437 eQTL**	[rs1864183]	C	T	49%	22	**HCC** [214]	Asian study	**MISSENSE** T212M	eQTL - Lung
						HNSCC RISK to develop laryngeal cancer [150] **NPC**, Bad Theragnosis (Radiotherapy) [222]	European study Asian study		
						NSCLC Poor survival[221] Theragnosis (Platinum)[67]	Asian study Asian study		
						PAGET DISEASE [200] Protective	European study		
						TUBERCULOSIS [223] ↑ IL8	Asian study		
	[rs3734114]	T	C	21%	36	**TUBERCULOSIS** [222] ↑ IL8	Asian study	Missense	(eQTL)
	[rs4703533]	C	G	39%	4	**HNSCC NPC** [222] Bad Theragnosis (Radiotherapy)	Asian study	Intronic	
	[rs10036653]	A	T	20%	1	**NSCLC** [224] ↓ RISK of brain metastasis **NSCLC EGFR*** [196] Theragnosis (Gefitinib) ↓ acquired resistance ↑ PFS ↑ OS	Asian study Asian study	5' *ATG10* ↑ OCT4 binding [196]	eQTL - Lung
	[rs10514231]	C	T	66%	1	**BREAST CANCER** [220] Protective **NSCLC** [221] Poor survival **HCC** [214] Protective **HNSCC NPC** [222] Bad Theragnosis (radiotherapy)	Asian study Asian study Asian study Asian study	Intronic	eQTL - Lung, Breast ↑ exp[214]
	[rs9447453]	C	A	11%	1	**Delayed age of MENOPAUSE** [225]	American study	5' *ATG10*	N/A

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ATG10 **NM_001131028 16437 eQTL**	rs1485587	A	G	47%	18	**ALZHEIMER DISEASE** [226]	American study	Intronic	eQTL - Brain
	rs324913	T	C			**ECZEMA HERPETICUM** [227] ↓ IFNγ	American study	Intronic	eQTL -Skin
	rs891159	G	A	22%	43	**ALZHEIMER DISEASE** [226]	American study	Intronic	eQTL - Brain
	rs4703879	A	G						eQTL - Brain
	rs7707921	T	A			**BREAST CANCER** [228]	European study	Intronic	eQTL - Breast
	rs73134739	T	C			**BREAST CANCER** [229]	Asian study	Intronic	eQTL - Breast
ATG12 **NM_004707 268 eQTL**	rs26532	C	A	78%	1	**NSCLC** [224] RISK of brain metastasis	Asian study	Intronic	eQTL - Brain
	rs26537	T	C	40%	5	**HNSCC**[230] **HCC** [214]	Asian study Asian study	Intronic	eQTL ↑ exp [230]
	rs26538	C	T			**NSCLC EGFR*** [196] Bad prognosis ↓ PFS **LUNG FIBROSIS** [197] **x COAL** - Protective	Asian study Asian study	Intronic	eQTL - Lung ↓ exp [196]
	rs1058600	C	T	23%	12	**NSCLC** [231] Theragnosis (Radiotherapy)	American study	3'UTR	eQTL - Lung
ATG16L1 **NM_030803 2507 eQTL**	rs2289476	C	A	6%	25	**CROHN DISEASE** [41,232,233] **x SMOKING**	European and American studies	Intronic	(eQTL)
	rs4663402	A	T	4%	18	**HNSCC** [230] **HCC** [214]	Asian study Asian study	Intronic	eQTL - Esophagus
	rs4663421	G	C			♀ **ANKYLOSING SPONDYLITIS** [234]	Asian study	Intronic	(eQTL)
	rs6758317	C	T	18%	3			Intronic	(eQTL)
	rs6754677	G	A	62%	8	**CROHN DISEASE** [235]	Asian study	Intronic	(eQTL)
	rs78835907	G	A	5%	1	**PROSTATE CANCER** [236] Bad prognostic	Asian study	N/A	N/A

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ATG16L1 **NM_030803 2507 eQTL**	[rs2241880]	A	G	53%	71	**CARDIOVASCULAR DISEASE** [237] RISK in postmenopausal women	Caucasian study	**MISSENSE** T300A	eQTL - Colon Thyroid
						COPD [238] **x SMOKING -** Protective	European study		
						COLORECTAL CANCER ↓ RISK ↓ metastasis ↑ IFN (indep. of autophagy) [239] ↑ RISK[240]	European study		
						CROHN DISEASE [41,232,233,235,241] Complications after CD ileocolectomy **x SMOKING**	European, American and Asian studies		
						GASTRIC CANCER [109,242] **x *H. pylori* infection**	Asians-Oceanian study		
						HCC RISK-based surveillance in cirrhosis [243] Protective: **HEPATITIS B** virus-related HCC [244]	European study Asian study		
						HNSCC [150] Oral carcinoma	European study		
						NSCLC **EGFR***minor allele: ↑ RISK Bad prognosis [196] Protective ↓ RISK of brain metastasis [224,238]	Asian and European studies		
						MELANOMA [198] Younger age	World collaboration		
						PREMATURE DELIVERY [245]	European Study		
						THYROID CANCER [246] Protective	European study		
						UROPATHOGENIC *E. coli* [247] Protective	American study		
	[rs13005285]	T	G	66%	8	**PALMOPLANTAR PUSTULOSIS** [248]	European study	Intronic	eQTL - Skin

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
ATG16L2 **NM_033388 222 eQTL**	rs10751215	T	C	47%	33	**Metastatic ccRCC** [117] **NSCLC** [231] Theragnosis (Radiotherapy) ↑ATG16L2 ↓ autophagosome ↑ inflammation	European study American study	Intronic	(eQTL)
	rs10898880	C	A					5' *ATG16L2*	(eQTL) ↑ **exp**[231]
	rs11605818	A	G	2%	1	**SLE** [249]	European study	Intronic	N/A
	rs11235604	C	T	0% As 10%	1	**CROHN DISEASE** [250–252] Protective **SLE** [253–255] with IgA nephropathy or lupus nephritis **NSCLC EGFR*** [196] Bad Theragnosis (Gefitinib)	American and Asian studies American and Asian studies Asian study	**MISSENSE** R220W	N/A ↓ **exp**
	rs11235667	A	G	0% As 11%	1	**SLE** [255] POL2, YY1, and FOXa binding sites	American and Asian Study	3' *ATG16L2*	N/A
CHMP2B **NM_014043 7278 eQTL**	rs1002765	G	A	11%	1	**ATROPHIC GASTRITIS** [256] precancerous **x *H. pylori* infection** **GASTRIC CANCER** [256] **x *H. pylori* infection**	Asian study	Intronic ⚠ not specific MIR4795	(eQTL)
	rs63751126	A	C	0%	1	**ALS** [257]	European study	**MISSENSE** Q206H	Not found
	rs63751048	C	T	0%	1	**FRONTOTEMPORAL DEMENTIA** [258] protein-truncating mutation	European study	nonsense	Not found
	rs63750652	G	A,C	0%	1			splice acceptor	Not found
	rs63750653	G	T	0%	1			**MISSENSE** N148T	Not found
LAMP1 **NM_005561 74 eQTL**	rs9577229	C	T	0%	1	**TUBERCULOSIS** [223]	Asian and European study	**MISSENSE** A204V	(eQTL)
	rs12871648	A	C	33%	1	**PARKINSON DISEASE** [259]	American study	Intronic	N/A
LAMP2 **NM_015104 820 eQTL**	rs727504953	G	A	0%	1	**DANON DISEASE** [260] cardiomyopathy, skeletal myopathy, and intellectual disability	Asian study	**MISSENSE** G93R	Not found ↓ exp [260]

VI - *AUTOPHAGY RECEPTORS*

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
DRAM1 **NM_018370 4677 eQTL** **induces the formation of the phagophore by binding p62/SQSTM-1** [261,262]	rs7955890	C	T	11%	40	**NSCLC** [67] Theragnostic (Platinum) ↑ PFS	Asian study	Intronic	eQTL - Lung
	rs17032060	G	A	1%	24			3'UTR	N/A
	rs77694286	A	G			**TYPE 2 DIABETES** [263] higher protein intake	American and European study **GWA 91,114**	Intronic	(eQTL) ↓ exp [263]
SQSTM1 **NM_003900 339 eQTL**	rs104893941	C	T	1%	1	**ALZHEIMER DISEASE** [264] Early-onset Rare germinal SQSTM1 mutations are associated with neurodegenerative diseases, such as ALS-FTD [265], Parkinson disease, Alzheimer disease [264], and also Paget disease of bone, metabolic diseases, obesity, and insulin resistance [266,267]	European study	**MISSENSE** P308L	N/A
NBR1 **NM_005899 10,959 eQTL** **Not specific. Commun NBR1, NBR2, BRCA1**	rs17599948	G	C	6%	6	**PROSTATE CANCER** [268] progression to lethal cancer after radiotherapy **BREAST CANCER** [269] Rare BRCA1 3'UTR SNPs	American study American and European study	Intronic	eQTL – Breast, Prostate
CALCOCO2 **NM_005831 1311 eQTL**	rs8074034	A	G	49%	26	**GLIOBLASTOMA** [270] pediatric	American and European study	Intronic	eQTL- Brain
	rs2303015	T	C	4%	75	**BACTERIAL PERITONITIS** [271] (spontaneous) in patients with alcoholic cirrhosis **CROHN DISEASE** [272]	European study American and European study	**MISSENSE** V272A	eQTL - Colon

Gene	variant	Ref	Alt	EUR freq	LD	Disease	Populations	Funct Annot	eQTL GTEX
WDFY3 **NM_014991 347 eQTL**	rs76117213	G	A	1%	1	**ALZHEIMER DISEASE** [273]	American study	Intronic	N/A
	rs17009220	G	C	6%	1			Intronic	N/A
	Heterozygous, mostly ***de novo*** **variants in WDFY3** result in a mild **NEURODEVELOPMENTAL** delay (intellectual disability, autism/hyperactivity disorder) **and opposing effects on brain size** [274]**: Loss-of-function** variants (truncating and missense) causing **haploinsufficiency** lead to **MACROCEPHALY. In contrast,** variants in the PH-domain of WDFY3 leads to **MICROCEPHALY, via dysregulation of the WNT-pathway**. Proliferating cortical neural progenitors highly express WDFY3, further supporting a role for this molecule in the regulation of prenatal neurogenesis.								
OPTN **NM_021980 473 eQTL**	rs3829923	C	T	34%	3	**GLAUCOMA** [275]	European study	CCDC3	N/A
	rs10796021	TT	T,TC	44%	1			CCDC3	Not found
	rs7921853	T	G	51%	1			Intronic	(eQTL)
	rs765884	T	C	28%	13			Intronic	(eQTL)
	rs11258194	T	A	3%	2	**GLAUCOMA** [276] **DIABETIC RETINOPATHY** [277]	Asian study Middle East study	**MISSENSE** M98K	N/A
	rs2234968	G	A	26%	4	**GLAUCOMA** [276]	American and Asian studies	synonymous	(eQTL)
	rs10906308	G	A	22%	18	**GLAUCOMA** [278]	American study	Intronic	Not found
	rs825411	A	G	54%	4	**PAGET DISEASE** [279]	European and Oceanian study	Intronic	(eQTL)
	rs1561570	T	C	50%	3			Intronic	(eQTL)

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