

The participation of the intrinsically disordered regions of the bHLH-PAS transcription factors in disease development

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Supplementary Materials

Results of HuVarBase : Human Variants Database for:

hAHR, hAHR, hSIM1, hSIM2, hHIF2a, hNPAS4, hARNT2, hBMAL1

<https://www.iitm.ac.in/bioinfo/huvarbase/mas18srch.php>

red letters mark residues located in predicted as disordered regions

hAHR (Uniprot P35869)

No.	Gene Name	Protein Mutation	Disease / Tissue (COSMIC)	Database
1	AHR	R554K	soft_tissue, cancer	COSMIC Humsavar
2	AHR	V570I	thyroid, cancer	COSMIC Humsavar
3	AHR	R398Q	large_intestine, cancer	COSMIC
4	AHR	Q383H	oesophagus, cancer	COSMIC
5	AHR	R368C	stomach, cancer	COSMIC
6	AHR	N284H	large_intestine, cancer	COSMIC
7	AHR	E488K	kidney, cancer	COSMIC
8	AHR	T507I	large_intestine, cancer	COSMIC
9	AHR	T311I	skin, cancer	COSMIC
10	AHR	T199P	large_intestine, cancer	COSMIC
11	AHR	T141N	central_nervous_system, cancer	COSMIC
12	AHR	S733F	liver, cancer	COSMIC
13	AHR	R305K	large_intestine, cancer	COSMIC
14	AHR	Q150K	liver, cancer	COSMIC
15	AHR	P838S	large_intestine, cancer	COSMIC
16	AHR	P260L	large_intestine, cancer	COSMIC
17	AHR	P18L	liver, cancer	COSMIC
18	AHR	N505S	large_intestine, cancer	COSMIC
19	AHR	E169K	breast, cancer	COSMIC
20	AHR	D132N	lung, cancer	COSMIC

hAHRR (Uniprot A9YTQ3)

No.	Gene Name	Protein Mutation	Disease / Tissue (COSMIC)	Database
1	AHRR 	R491Q	prostate, cancer	ClinVar  COSMIC 
2	AHRR 	P189A	central_nervous_system, cancer	COSMIC  Humsavar 
3	AHRR 	T152M	large_intestine, cancer	COSMIC 
4	AHRR 	R285W	large_intestine, cancer	COSMIC 
5	AHRR 	D645H	prostate, cancer	COSMIC 
6	AHRR 	A301V	skin, cancer	COSMIC 
7	AHRR 	A112V	liver, cancer	COSMIC 
8	AHRR 	V553M	large_intestine, cancer	COSMIC 
9	AHRR 	V29M	large_intestine, cancer	COSMIC 
10	AHRR 	T524M	large_intestine, cancer	COSMIC 
11	AHRR 	T419I	large_intestine, cancer	COSMIC 
12	AHRR 	S63F	large_intestine, cancer	COSMIC 
13	AHRR 	S53G	stomach, cancer	COSMIC 
14	AHRR 	R491W	large_intestine, cancer	COSMIC 
15	AHRR 	R485W	large_intestine, cancer	COSMIC 
16	AHRR 	R230C	large_intestine, cancer	COSMIC 
17	AHRR 	Q88R	large_intestine, cancer	COSMIC 
18	AHRR 	P283S	skin, cancer	COSMIC 
19	AHRR 	P102S	large_intestine, cancer	COSMIC 
20	AHRR 	I226V	large_intestine, cancer	COSMIC 
21	AHRR 	G612S	oesophagus, cancer	COSMIC 
22	AHRR 	G494S	large_intestine, cancer	COSMIC 
23	AHRR 	G427E	skin, cancer	COSMIC 
24	AHRR 	C545F	liver, cancer	COSMIC 
25	AHRR 	A96V	large_intestine, cancer	COSMIC 
26	AHRR 	A674S	liver, cancer	COSMIC 
27	AHRR 	A371T	endometrium, cancer	COSMIC 
28	AHRR 	A300T	large_intestine, cancer	COSMIC 

SIM1 (Uniprot P81133)

No.	Gene Name	Protein Mutation	Disease / Tissue (COSMIC)	Database
1	SIM1 	P352T	breast, cancer	1000 Genomes  (data @ Ensembl ) ClinVar  COSMIC  Humsavar 
2	SIM1 	A371V	large_intestine, cancer	1000 Genomes  (data @ Ensembl ) ClinVar  COSMIC  Humsavar 
3	SIM1 	H268Q	Schaaf-yang syndrome	ClinVar 
4	SIM1 	Q704L	Schaaf-yang syndrome	ClinVar 
5	SIM1 	E725K	oesophagus, cancer	ClinVar  COSMIC 
6	SIM1 	G408R	haematopoietic_and_lymphoid_tissue, cancer	COSMIC 
7	SIM1 	E399K	skin, cancer	COSMIC 
8	SIM1 	T481M	haematopoietic_and_lymphoid_tissue, cancer	COSMIC 
9	SIM1 	R192H	lung, cancer	COSMIC 
10	SIM1 	H559Q	liver, cancer	COSMIC 
11	SIM1 	C472W	large_intestine, cancer	COSMIC 
12	SIM1 	S454L	skin, cancer	COSMIC 
13	SIM1 	R471Q	skin, cancer	COSMIC 
14	SIM1 	H394Y	skin, cancer	COSMIC 
15	SIM1 	G392R	lung, cancer	COSMIC 
16	SIM1 	E224K	skin, cancer	COSMIC 
17	SIM1 	D424N	skin, cancer	COSMIC 
18	SIM1 	A494T	breast, cancer	COSMIC 
19	SIM1 	A311V	large_intestine, cancer	COSMIC 
20	SIM1 	V326I	stomach, cancer	COSMIC 
21	SIM1 	V222I	stomach, cancer	COSMIC 
22	SIM1 	V213M	cervix, cancer	COSMIC 
23	SIM1 	T292N	liver, cancer	COSMIC 
24	SIM1 	S701C	lung, cancer	COSMIC 
25	SIM1 	S680L	prostate, cancer	COSMIC 
26	SIM1 	S663L	large_intestine, cancer	COSMIC 
27	SIM1 	S603F	skin, cancer	COSMIC 
28	SIM1 	S541L	stomach, cancer	COSMIC 

29	SIM1	S428F	skin, cancer	COSMIC
30	SIM1	S31L	urinary_tract, cancer	COSMIC
31	SIM1	S309G	upper_aerodigestive_tract, cancer	COSMIC
32	SIM1	R665C	skin, cancer	COSMIC
33	SIM1	R657W	large_intestine, cancer	COSMIC
34	SIM1	R550C	skin, cancer	COSMIC
35	SIM1	R548Q	large_intestine, cancer	COSMIC
36	SIM1	R493C	skin, cancer	COSMIC
37	SIM1	R192C	large_intestine, cancer	COSMIC
38	SIM1	R10W	large_intestine, cancer	COSMIC
39	SIM1	Q704H	liver, cancer	COSMIC
40	SIM1	Q36P	lung, cancer	COSMIC
41	SIM1	P661L	skin, cancer	COSMIC
42	SIM1	P588L	skin, cancer	COSMIC
43	SIM1	P539R	central_nervous_system, cancer	COSMIC
44	SIM1	N650Y	lung, cancer	COSMIC
45	SIM1	L217P	large_intestine, cancer	COSMIC
46	SIM1	H402Y	skin, cancer	COSMIC
47	SIM1	H268Y	skin, cancer	COSMIC
48	SIM1	G65D	upper_aerodigestive_tract, cancer	COSMIC
49	SIM1	L217P	large_intestine, cancer	COSMIC
50	SIM1	G448C	liver, cancer	COSMIC
51	SIM1	G303S	stomach, cancer	COSMIC
52	SIM1	G271S	oesophagus, cancer	COSMIC
53	SIM1	E530K	lung, cancer	COSMIC
54	SIM1	E3D	large_intestine, cancer	COSMIC
55	SIM1	E396D	large_intestine, cancer	COSMIC
56	SIM1	E155K	skin, cancer	COSMIC
57	SIM1	D74Y	urinary_tract, cancer	COSMIC
58	SIM1	A570G	lung, cancer	COSMIC
59	SIM1	A435T	stomach, cancer	COSMIC
60	SIM1	A432T	large_intestine, cancer	COSMIC
61	SIM1	A236T	large_intestine, cancer	COSMIC

SIM2 (Uniprot Q14190)

No.	Gene Name	Protein Mutation	Disease / Tissue (COSMIC)	Database
1	SIM2 	L483M	thyroid, cancer	COSMIC  Humsavar 
2	SIM2 	M377I	skin, cancer	COSMIC 
3	SIM2 	D273N	skin, cancer	COSMIC 
4	SIM2 	R318H	prostate, cancer	COSMIC 
5	SIM2 	F56L	liver, cancer	COSMIC 
6	SIM2 	A70T	liver, cancer	COSMIC 
7	SIM2 	A63V	large_intestine, cancer	COSMIC 
8	SIM2 	A280T	large_intestine, cancer	COSMIC 
9	SIM2 	Q469P	lung, cancer	COSMIC 
10	SIM2 	Y194H	skin, cancer	COSMIC 
11	SIM2 	Y125H	liver, cancer	COSMIC 
12	SIM2 	V92F	skin, cancer	COSMIC 
13	SIM2 	V76I	thyroid, cancer	COSMIC 
14	SIM2 	V326M	large_intestine, cancer	COSMIC 
15	SIM2 	V326G	thyroid, cancer	COSMIC 
16	SIM2 	V313A	lung, cancer	COSMIC 
17	SIM2 	V211G	breast, cancer	COSMIC 
18	SIM2 	T646P	lung, cancer	COSMIC 
19	SIM2 	T433M	large_intestine, cancer	COSMIC 
20	SIM2 	T253M	breast, cancer	COSMIC 
21	SIM2 	T223I	skin, cancer	COSMIC 
22	SIM2 	T120M	central_nervous_system, cancer	COSMIC 
23	SIM2 	S503Y	cervix, cancer	COSMIC 
24	SIM2 	S502W	upper_aerodigestive_tract, cancer	COSMIC 
25	SIM2 	S355F	lung, cancer	COSMIC 
26	SIM2 	S343Y	lung, cancer	COSMIC 
27	SIM2 	S199Y	bone, cancer	COSMIC 
28	SIM2 	R44G	kidney, cancer	COSMIC 
29	SIM2 	R318L	oesophagus, cancer	COSMIC 
30	SIM2 	R278C	liver, cancer	COSMIC 
31	SIM2 	P57S	skin, cancer	COSMIC 
32	SIM2 	P448S	skin, cancer	COSMIC 
33	SIM2 	P385H	lung, cancer	COSMIC 
34	SIM2 	P145L	prostate, cancer	COSMIC 
35	SIM2 	M231I	liver, cancer	COSMIC 
36	SIM2 	M164I	skin, cancer	COSMIC 
37	SIM2 	L283V	prostate, cancer	COSMIC 

38	SIM2	L240P	thyroid, cancer	COSMIC
39	SIM2	L168F	large_intestine, cancer	COSMIC
40	SIM2	K368N	cervix, cancer	COSMIC
41	SIM2	K190N	endometrium, cancer	COSMIC
42	SIM2	I124M	stomach, cancer	COSMIC
43	SIM2	H267Y	skin, cancer	COSMIC
44	SIM2	H147Y	thyroid, cancer	COSMIC
45	SIM2	G303S	pancreas, cancer	COSMIC
46	SIM2	G271D	large_intestine, cancer	COSMIC
47	SIM2	G254R	skin, cancer	COSMIC
48	SIM2	G174S	liver, cancer	COSMIC
49	SIM2	F454S	skin, cancer	COSMIC
50	SIM2	F394S	liver, cancer	COSMIC
51	SIM2	E345K	skin, cancer	COSMIC
52	SIM2	E339K	skin, cancer	COSMIC
53	SIM2	E262K	skin, cancer	COSMIC
54	SIM2	E106K	large_intestine, cancer	COSMIC
55	SIM2	D450N	skin, cancer	COSMIC
56	SIM2	D246N	skin, cancer	COSMIC
57	SIM2	D239Y	liver, cancer	COSMIC
58	SIM2	D202N	large_intestine, cancer	COSMIC
59	SIM2	D134N	lung, cancer	COSMIC
60	SIM2	C489G	fallopian_tube, cancer	COSMIC
61	SIM2	C324Y	thyroid, cancer	COSMIC
62	SIM2	C324F	liver, cancer	COSMIC
63	SIM2	A40V	lung, cancer	COSMIC
64	SIM2	A350S	haematopoietic_and_lymphoid_tissue, cancer	COSMIC
65	SIM2	A311V	bone, cancer	COSMIC
66	SIM2	A221T	stomach, cancer	COSMIC
67	SIM2	A212V	large_intestine, cancer	COSMIC
68	SIM2	A169V	large_intestine, cancer	COSMIC
69	SIM2	A108T	liver, cancer	COSMIC

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HIF-2 α (EPAS1, Uniprot Q99814)

No.	Gene Name	Protein Mutation	Disease / Tissue (COSMIC)	Database
1	EPAS1 	G537R	Erythrocytosis, familial, 4	ClinVar  Humsavar  SwissVar  (variant page )
2	EPAS1 	G537W	Erythrocytosis, familial, 4	ClinVar  Humsavar  SwissVar  (variant page )
3	EPAS1 	M535V	Erythrocytosis, familial, 4	ClinVar  Humsavar  SwissVar  (variant page )
4	EPAS1 	P785T	Familial erythrocytosis	1000 Genomes  (data @ Ensembl ) ClinVar  Humsavar 
5	EPAS1 	T766P	pancreas, cancer	1000 Genomes  (data @ Ensembl ) ClinVar  COSMIC  Humsavar 
6	EPAS1 	A410T	Familial erythrocytosis	1000 Genomes  (data @ Ensembl ) ClinVar 
7	EPAS1 	E832D	Familial erythrocytosis	1000 Genomes  (data @ Ensembl ) ClinVar 
8	EPAS1 	F608L	Familial erythrocytosis	ClinVar 
9	EPAS1 	I789V	Familial erythrocytosis	ClinVar 
10	EPAS1 	Q294H	Familial erythrocytosis	ClinVar 
11	EPAS1 	R798G	Familial erythrocytosis	1000 Genomes  (data @ Ensembl ) ClinVar 
12	EPAS1 	R825Q	Familial erythrocytosis	ClinVar 
13	EPAS1 	S49Y	Familial erythrocytosis	ClinVar 
14	EPAS1 	S703A	Familial erythrocytosis	ClinVar 
15	EPAS1 	F540L	erythrocytosis, familial, 4	Humsavar  SwissVar  (variant page )
16	EPAS1 	M535T	erythrocytosis, familial, 4	Humsavar  SwissVar  (variant page )
17	EPAS1 	A530V	autonomic_ganglia, cancer	COSMIC 
18	EPAS1 	P531L	adrenal_gland, cancer	COSMIC 
19	EPAS1 	Y532C	adrenal_gland, cancer	COSMIC 
20	EPAS1 	L529P	autonomic_ganglia, cancer	COSMIC 
21	EPAS1 	P531S	adrenal_gland, cancer	COSMIC 
22	EPAS1 	A530T	pancreas, cancer	COSMIC 

23	EPAS1 	A530E	autonomic_ganglia, cancer	COSMIC 
24	EPAS1 	M507T	prostate, cancer	COSMIC 
25	EPAS1 	G314E	haematopoietic_and_lymphoid_tissue, cancer	COSMIC 
26	EPAS1 	E82K	haematopoietic_and_lymphoid_tissue, cancer	COSMIC 
27	EPAS1 	E279V	liver, cancer	COSMIC 
28	EPAS1 	D753E	oesophagus, cancer	COSMIC 
29	EPAS1 	D539Y	autonomic_ganglia, cancer	COSMIC 
30	EPAS1 	Y489H	large_intestine, cancer	COSMIC 
31	EPAS1 	V317M	large_intestine, cancer	COSMIC 
32	EPAS1 	T31M	large_intestine, cancer	COSMIC 
33	EPAS1 	S72L	lung, cancer	COSMIC 
34	EPAS1 	S723N	ovary, cancer	COSMIC 
35	EPAS1 	S672Y	large_intestine, cancer	COSMIC 
36	EPAS1 	S55F	breast, cancer	COSMIC 
37	EPAS1 	S474T	stomach, cancer	COSMIC 
38	EPAS1 	S372N	large_intestine, cancer	COSMIC 
39	EPAS1 	S355F	skin, cancer	COSMIC 
40	EPAS1 	S276L	large_intestine, cancer	COSMIC 
41	EPAS1 	R710Q	lung, cancer	COSMIC 
42	EPAS1 	R144C	upper_aerodigestive_tract, cancer	COSMIC 
43	EPAS1 	P727L	urinary_tract, cancer	COSMIC 
44	EPAS1 	N768T	large_intestine, cancer	COSMIC 
45	EPAS1 	L542R	liver, cancer	COSMIC 
46	EPAS1 	H248N	large_intestine, cancer	COSMIC 
47	EPAS1 	A94T	liver, cancer	COSMIC 

NPAS4 (Uniprot Q8IUM7)

No.	Gene Name	Protein Mutation	Disease / Tissue (COSMIC)	Database
1	NPAS4 	T587M	central_nervous_system, cancer	COSMIC  Humsavar 
2	NPAS4 	R220H	large_intestine, cancer	COSMIC 
3	NPAS4 	D419N	skin, cancer	COSMIC 
4	NPAS4 	T708M	stomach, cancer	COSMIC 
5	NPAS4 	R595W	kidney, cancer	COSMIC 
6	NPAS4 	R150L	liver, cancer	COSMIC 
7	NPAS4 	S747F	liver, cancer	COSMIC 
8	NPAS4 	S544N	skin, cancer	COSMIC 
9	NPAS4 	S453C	upper_aerodigestive_tract, cancer	COSMIC 
10	NPAS4 	R392Q	NS, cancer	COSMIC 
11	NPAS4 	R159C	large_intestine, cancer	COSMIC 
12	NPAS4 	L455F	skin, cancer	COSMIC 
13	NPAS4 	L322I	large_intestine, cancer	COSMIC 
14	NPAS4 	G83S	large_intestine, cancer	COSMIC 
15	NPAS4 	G566E	NS, cancer	COSMIC 
16	NPAS4 	E725K	skin, cancer	COSMIC 
17	NPAS4 	D730N	skin, cancer	COSMIC 
18	NPAS4 	D121N	large_intestine, cancer	COSMIC 
19	NPAS4 	A63V	stomach, cancer	COSMIC 
20	NPAS4 	A592V	oesophagus, cancer	COSMIC 
21	NPAS4 	V710M	oesophagus, cancer	COSMIC 
22	NPAS4 	V167M	breast, cancer	COSMIC 
23	NPAS4 	T558I	skin, cancer	COSMIC 
24	NPAS4 	S683I	liver, cancer	COSMIC 
25	NPAS4 	S493L	thyroid, cancer	COSMIC 
26	NPAS4 	S219N	large_intestine, cancer	COSMIC 
27	NPAS4 	S156F	NS, cancer	COSMIC 
28	NPAS4 	R634H	pancreas, cancer	COSMIC 
29	NPAS4 	R51H	large_intestine, cancer	COSMIC 
30	NPAS4 	R200H	breast, cancer	COSMIC 
31	NPAS4 	R172Q	large_intestine, cancer	COSMIC 
32	NPAS4 	R145C	skin, cancer	COSMIC 
33	NPAS4 	R132H	breast, cancer	COSMIC 
34	NPAS4 	Q629H	cervix, cancer	COSMIC 
35	NPAS4 	Q547H	liver, cancer	COSMIC 
36	NPAS4 	Q469H	upper_aerodigestive_tract, cancer	COSMIC 
37	NPAS4 	Q332K	liver, cancer	COSMIC 

38	NPAS4 	P82H	skin, cancer	COSMIC 
39	NPAS4 	P679L	liver, cancer	COSMIC 
40	NPAS4 	P597S	endometrium, cancer	COSMIC 
41	NPAS4 	P533S	skin, cancer	COSMIC 
42	NPAS4 	P533L	skin, cancer	COSMIC 
43	NPAS4 	P405L	liver, cancer	COSMIC 
44	NPAS4 	P199H	large_intestine, cancer	COSMIC 
45	NPAS4 	P194S	skin, cancer	COSMIC 
46	NPAS4 	P194L	liver, cancer	COSMIC 
47	NPAS4 	L351I	large_intestine, cancer	COSMIC 
48	NPAS4 	I639V	liver, cancer	COSMIC 
49	NPAS4 	I236V	NS, cancer	COSMIC 
50	NPAS4 	G204D	central_nervous_system, cancer	COSMIC 
51	NPAS4 	E724K	bone, cancer	COSMIC 
52	NPAS4 	E628G	breast, cancer	COSMIC 
53	NPAS4 	D716N	skin, cancer	COSMIC 
54	NPAS4 	D647N	liver, cancer	COSMIC 
55	NPAS4 	A8T	large_intestine, cancer	COSMIC 
56	NPAS4 	A210V	liver, cancer	COSMIC 
57	NPAS4 	A175T	oesophagus, cancer	COSMIC 

ARNT2 (Uniprot Q9HBZ2)

No.	Gene Name	Protein Mutation	Disease / Tissue (COSMIC)	Database
1	ARNT2 	R47C	large_intestine, cancer	COSMIC 
2	ARNT2 	V167I	liver, cancer	COSMIC 
3	ARNT2 	S458L	skin, cancer	COSMIC 
4	ARNT2 	R76W	endometrium, cancer	COSMIC 
5	ARNT2 	R404C	skin, cancer	COSMIC 
6	ARNT2 	P529S	skin, cancer	COSMIC 
7	ARNT2 	Y430N	central_nervous_system, cancer	COSMIC 
8	ARNT2 	V683L	lung, cancer	COSMIC 
9	ARNT2 	V110I	large_intestine, cancer	COSMIC 
10	ARNT2 	T602M	large_intestine, cancer	COSMIC 
11	ARNT2 	S343F	upper_aerodigestive_tract, cancer	COSMIC 
12	ARNT2 	S332L	pancreas, cancer	COSMIC 
13	ARNT2 	R652Q	large_intestine, cancer	COSMIC 
14	ARNT2 	R240K	large_intestine, cancer	COSMIC 
15	ARNT2 	R209Q	upper_aerodigestive_tract, cancer	COSMIC 
16	ARNT2 	P579S	large_intestine, cancer	COSMIC 
17	ARNT2 	P423S	skin, cancer	COSMIC 
18	ARNT2 	P269S	pancreas, cancer	COSMIC 
19	ARNT2 	M328I	lung, cancer	COSMIC 
20	ARNT2 	I105V	central_nervous_system, cancer	COSMIC 
21	ARNT2 	H543R	haematopoietic_and_lymphoid_tissue, cancer	COSMIC 
22	ARNT2 	G710A	liver, cancer	COSMIC 
23	ARNT2 	G31R	stomach, cancer	COSMIC 
24	ARNT2 	E72K	stomach, cancer	COSMIC 
25	ARNT2 	D344N	large_intestine, cancer	COSMIC 
26	ARNT2 	D344G	endometrium, cancer	COSMIC 
27	ARNT2 	D191G	liver, cancer	COSMIC 
28	ARNT2 	A28V	large_intestine, cancer	COSMIC 
29	ARNT2 	A25T	lung, cancer	COSMIC 

BMAL1 (Uniprot O00327)

No.	Gene Name	Protein Mutation	Disease / Tissue (COSMIC)	Database
1	ARNTL 	R84H	stomach, cancer	COSMIC 
2	ARNTL 	V260A	large_intestine, cancer	COSMIC 
3	ARNTL 	R83Q	liver, cancer	COSMIC 
4	ARNTL 	R37C	large_intestine, cancer	COSMIC 
5	ARNTL 	R244Q	large_intestine, cancer	COSMIC 
6	ARNTL 	R166G	breast, cancer	COSMIC 
7	ARNTL 	P234H	skin, cancer	COSMIC 
8	ARNTL 	D145N	large_intestine, cancer	COSMIC 
9	ARNTL 	A345T	liver, cancer	COSMIC 
10	ARNTL 	R37H	large_intestine, cancer	COSMIC 
11	ARNTL 	R238Q	large_intestine, cancer	COSMIC 
12	ARNTL 	P292T	pancreas, cancer	COSMIC 
13	ARNTL 	V162I	biliary_tract, cancer	COSMIC 
14	ARNTL 	T224S	stomach, cancer	COSMIC 
15	ARNTL 	T140S	breast, cancer	COSMIC 
16	ARNTL 	S90I	haematopoietic_and_lymphoid_tissue, cancer	COSMIC 
17	ARNTL 	S372L	breast, cancer	COSMIC 
18	ARNTL 	S27Y	large_intestine, cancer	COSMIC 
19	ARNTL 	S246C	cervix, cancer	COSMIC 
20	ARNTL 	R85Q	ovary, cancer	COSMIC 
21	ARNTL 	Q4L	liver, cancer	COSMIC 
22	ARNTL 	Q190E	upper_aerodigestive_tract, cancer	COSMIC 
23	ARNTL 	P198L	skin, cancer	COSMIC 
24	ARNTL 	M88I	NS, cancer	COSMIC 
25	ARNTL 	I80F	thyroid, cancer	COSMIC 
26	ARNTL 	H66P	thyroid, cancer	COSMIC 
27	ARNTL 	E65K	genital_tract, cancer	COSMIC 
28	ARNTL 	E62Q	oesophagus, cancer	COSMIC 
29	ARNTL 	E375G	large_intestine, cancer	COSMIC 
30	ARNTL 	D299Y	liver, cancer	COSMIC 
31	ARNTL 	D22N	large_intestine, cancer	COSMIC 
32	ARNTL 	D145E	pancreas, cancer	COSMIC 
33	ARNTL 	D110Y	large_intestine, cancer	COSMIC 
34	ARNTL 	C249R	thyroid, cancer	COSMIC 
35	ARNTL 	A104T	central_nervous_system, cancer	COSMIC 