REPORT #60000

Below we describe the meanings of the symbols/abbreviations used in the report.

- **ICDM name:** Standardized disease names taken in accordance with International Statistical Classifcation of Diseases and Related Health Problems (ICD). This is the name of disease that your primary physician will recognize and use to make decisions about your health.
- **ICDM ID:** Corresponding disease names ID taken from International Statistical Classifcation of Diseases and Related Health Problems (ICD). This is the number (the tag) of disease listed in your primary physician booklet and used by your health insurance provider.
- **ICDM score:** Disease name matching score using ICDM (maximum 100). This is a measure of confidence of matching scientific descriptors and ICD names. The large number(but smaller than 100) indicates high confidence.
- **MESH name:** Standardized disease names taken in accordance with Medical Subject Headings (MESH). This is reported simply for completeness and general customer should not pay attention to it. Interested individuals should contact Predicagen LLC at support@predicagen.com for more details.
- **MESH ID:** Corresponding disease names ID taken from Medical Subject Headings (MESH). Similarly as above.
- **MESH score:** Disease name matching score using MESH(maximum 100). High score (but smaller than 100) indicates high confidence of predictions.
- **Phenotype:** The set of observable characteristics of corresponding disease. This is scientific description of effect of the DNA variant(s) on functionality of the corresponding biological macromolecules. If you want to know more how your DNA variants affect the corresponding macromolecules functionality at molecular level, you should contact us at support@predicagen.com.
- **RS ID:** The genetic variants RS ID. This is a list of DNA variants associated with the corresponding disease. More RS numbers associated with a disease indicate higher probability that you are at risk.

Begin of Table

ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
3-methylglutaconic aciduria	e71111	89	3-methylglutaconic aciduria, type i	218476	94	3-Methylglutaconic aciduria type 3	200203460 200203460 121913502 121913502 28937899 28937899
abdominal aortic aneurysm, ruptured	i713	85	aortic aneurysm, abdominal	219957	85	sporadic abdominal aortic aneurysm	2230267
abnormal brain scan	r9402	72	fibrinogen baltimore i	70352	91	FIBRINOGEN BALTIMORE 2	4220
abnormal glucose complicating pregnancy	o99810	71	intrahepatic cholestasis of pregnancy	197118	97	Cholestasis, intrahepatic, of pregnancy 3	45575636 72552778 72552780
abnormal head movements	r250	70	hb castilla	34750	75	G6PD CASTILLA	1799971 35291591 33935780 35854892 34980264 35492035 34095019 34188626 34139813 35871407 34022507 35067717 36081208 33972927 33972927 35068498 35351128 35819837 33952147 34173382 35286210 34362537 36008922 36008922 34151786 34933455 34974709 33935983 34037627 35262412 35262412 34378160 34378160 34571024 33950507 63749918 34866629 35802118 34387455 35906307 34149886 35848600 33956485 35849348 34802738 36078803 34536353 33971270 35329985 28933076 28933077 34933313 35913713 34977235 35166721 35152987 35395083 34093840 34012192 35433207 63749797 41330850 35020253 34127117 34435255 35481866 34915311 35746147 34017450 41475844 33974602 34532478 35977759 35700518 41404150 35885783 35654328 35957832 34907654 34438981 34263826 34807671 1802959 41341344 63750122 41510746 41323248 63749997 34440919 34440919 41417548 41361546 41393644 41321345 36030576 28928884 35059618 35974739 35166834 34011123 34849179 35723200 1050829 1050828
absence of iris	q131	65	epilepsy, childhood absence, susceptibility to, 4	221132	69	Colorectal cancer, susceptibility to	1801155 1042714 25409 3751664 72552322 17879961
absolute glaucoma, left eye	h44512	65	glaucoma 1, open angle, e	219312	100	Glaucoma 1, open angle, e	28939688
acanthosis nigricans	l83	100	insulin resistance	198576	100	Insulin resistance	60662302 5744168 1801278 1044498 1169288
acetonuria	r824	73	alkaptonuria	193518	100	Alkaptonuria	28942100
achondrogenesis	q770	61	alcohol dehydrogenase	57816	72	Alcohol dependence	2066702 1229984 1693482 671
achondroplasia	q774	100	pseudoachondroplasia	197023	98	Pseudoachondroaplasia	28931614 28931614 28933068 28933068 28936669
achromatopsia	h5351	93	achromatopsia 1	197192	93	Achromatopsia 6	147118493 200311463
acidosis	e872	84	sarcoidosis, early-onset	218972	100	Sarcoidosis, early-onset	5743291
acquired coagulation factor deficiency	d684	73	vitamin k-dependent clotting factors, combined deficiency of, 1	219639	98	Vitamin k-dependent clotting factors, combined deficiency of, 2	28928872 72547528
acquired ichthyosis	l850	63	ichthyosis, cyclic, with epidermolytic hyperkeratosis	219400	100	Ichthyosis, cyclic, with epidermolytic hyperkeratosis	57837128 61218439
acquired keratosis follicularis	l110	64	thyroid cancer, follicular	225806	100	Thyroid cancer, follicular	11554290 28933406
acquired keratosis follicularis	l110	64	thyroid cancer, follicular	225806	100	Thyroid cancer, follicular	
activated protein c resistance	d6851	63	cdc2-selective inhibitory kinase, rat	156346	62	Selective serotonin reuptake inhibitors response - Efficacy	6025 4713916 8177374 7997012 1800566 13306278
activity, running	y9302	61	actinin, alpha 3, human	115953	73	ACTININ, ALPHA-3 POLYMORPHISM	1815739
acute cholecystitis	k810	81	cholecystitis	197835	100	Cholecystitis	45575636 72552778 58238559
acute inflammatory disease of uterus	n710	62	interferon-gamma-responsive transcript-1 protein, human	111077	59	peginterferon alfa-2b and ribavirin response - Toxicity/ADR	11209026 2241880 10065172 12979860 12979860 8099917 8099917 8099917 1127354 1127354
acute intermittent (hepatic) porphyria	e8021	88	porphyria, acute intermittent	220977	100	Acute intermittent porphyria	34413634
acute lymphoblastic leukemia, in relapse	c9102	80	aieop acute lymphoblastic leukemia protocol	79478	72	Leukemia, acute lymphoblastic, susceptibility to	61754966

Continuation of Table

ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
acute myeloblastic leukemia, in relapse	c9202	70	leukemia, myeloid, acute	218692	100	Acute myeloid leukemia	77375493 28931590
acute prostatitis	n410	65	rosuvastatin lactone	209894	72	rosuvastatin response - Other	4693075 2231142 4149056
acute vaginitis	n760	60	cancer vaccines	3879	76	Ovarian cancer	3092989 9567552 1799943 1799943 1799943 206120 4987046 2126042 2126042 28897705 766173 766173 11571640 28897706 144848 144848 41293475 56403624 28897708 28897709 28897710 11571653 28897715 2227943 28897717 28897718 28897718 1799944 55638633 41293477 28897719 1801406 1801406 28897720 28897722 28897723 28897725 55969723 28897726 28897727 28897728 41293491 28897731 56087561 28897735 11571657 41293497 41293497 41293497 55875643 55875643 4987117 28897737 1799954 56191579 56191579 34309943 35029074 35029074 55953736 28897742 206079 11571684 206081 11571686 9943876 55977008 4986859 28897744 9534262 28897745 28897746 28897747 41293511 41293513 41293513 41293513 28897749 28897749 28897750 9634672 28897751 11571747 28897753 4942486 28897754 28897754 11571769 28897755 28897756 45580035 11571789 206147 11571805 41293521 28897758 28897758 80359205 28897759 61757642 517118 11571831 11571831 11571833 28897761 28897761 15869 28363284 8176320 8176319 8176318 41293465 8176316 1800751 28897698 28897698 3092988 8176289 45553935 45553935 8176273 8176265 41293461 41293461 3092994 45444999 28897696 28897696 55770810 8176258 1799967 1800726 1799966 1799966 28897693 56158747 28897692 28897691 28897691 55815649 1800744 8176199 3737559 4986849 41293455 41293455 1060915 28897690 799916 28897689 55639854 28897686 28897686 55930959 55725337 62625307 28897685 16942 55909400 4986852 16941 16941 1800704 56321129 799917 799917 799917 41286300 28897684 1800709 28897683 56082113 28897682 62625306 62625306 16940 41286296 4986846 4986845 4986844 4986850 4986850 28897680 1800064 1800064 56039126 56012641 28897677 28897676 28897676 62625300 56128296 1799950 8176153 28897675 1799965 34545365 799923 799912 28897672 28897672 8176109 8176104 8176103 1800062 1800062 8176092 8176091 3765640 3092986 11655505 4793204
acute vaginitis	n760	60	cancer vaccines	3879	76	Ovarian cancer	
acute vaginitis	n760	60	cancer vaccines	3879	76	Ovarian cancer	
agalactia	o923	63	1-deoxygalactonojirimycin	90673	81	Deoxygalactonojirimycin response	28935490
agnosia	r481	71	heterotropan	73016	87	Heterotopia	28937880 28935470
alopecia universalis	l631	100	alopecia universalis congenita	198075	100	Alopecia universalis congenita	7014851
alpers disease	g3181	80	cap myopathy	231507	80	Myopathy	57077886 28928903 58922911 28928900 59270054 41313880 60864230 59026483 61195471 28933092 61295588 60682848 57207746 59332535 11264444 56816490 56771886 56851164 59301204 60458016 61672878 534807 58932704 57629361 57629361 60934003 56984562 56984562 61444459 60890628 45466197 63750197 3766871 34967813 6716782 201273719 28933405 62636495 60798368 62636491 57496341 57639980 62636492 58898021 57955682 73991549 28936685 63750743 41261344 45546039 45620037 41276525 34768413 28938173 28937594 62541771 201103536 147622517 202196166 201306690 71579353 1417635 36212066 35078470 2856655 36211723 11570082 34580776 2228387 35404804 61001398 58386780 193922674 1046116 201754030 28928909 201728041 28933098 45464193 45544633 45451303 45478699 3218716 3218713 45516091 45614536 45578741 62635764 59628143 57815192 57815192 58008462 58645997 58645997 62636501 60825166 28932769 59985777 58536923 61497286 59565950 59565950 59565950 60269890 56679084 57661783 59568967 61622935 61622935 60449251 59285727 59285727 59285727 59793293 59793293 57590980 57120761 57120761 61060395 61060395 61060395 60343255 60095124 2230234 76992529 200563280 6060980 199896820 28937598 28937868 28935490 3134560
alpha-1-antitrypsin deficiency	e8801	100	alpha-1 antitrypsin deficiency	220634	100	Alpha-1-antitrypsin deficiency	61761869 28929474 17580 28929470 199422211
alpha thalassemia	d560	100	alpha-thalassemia	199118	87	Alpha plus thalassemia	34690599 34451549 11549407 35004220 35724775 33915217 33971440 33986703 35699606 35497102 34598529 33944208 63751457
alveolar proteinosis	j8401	63	protein-tyrosine kinase xyk, xenopus	116752	63	Deep venous thrombosis, protection against	2814778 887829 4693075 2231142 72552713 2231137 1800449 351855 4713916 72658152 4244285 5219 1954787 4149056 62642055 7997012 1719247 1346268 17822931 6048
alzheimer's disease with early onset	g300	87	alzheimer disease early onset type 3	197691	77	Alzheimer disease, early-onset, susceptibility to	63749824 63750815 63751141 63751037 63750590 63750577 63751144 63751068 63750526 63751163 63751320 63751229 63751235 63751223 1799990
alzheimer's disease with late onset	g301	86	alzheimer disease 2	197689	100	Alzheimer disease 2	2227564 429358
amyotrophic lateral sclerosis	g1221	100	amyotrophic lateral sclerosis, juvenile, with dementia	220447	63	Amyotrophic lateral sclerosis-parkinsonism/dementia complex 1, susceptibility to	63750818 58599399 8042919
anal prolapse	k622	62	nodal protein	188185	64	Nodal rhythm	45620037
anal prolapse	k622	62	nodal protein	188185	64	Nodal rhythm	
anencephaly	q000	100	neural tube defects, folate-sensitive	197535	80	Neural tube defects, folate-sensitive, susceptibility to	1801394 2236225
anhidrosis	l744	67	fluticasone propionate-17-carboxylic acid	208261	67	fluticasone propionate response - Efficacy	1876828
anisakiasis	b810	67	glutamic pyruvate transaminase 2, human	13758	81	GLUTAMIC PYRUVATE TRANSAMINASE POLYMORPHISM	1063739
ankylosis, right foot	m24674	80	tooth ankylosis	197503	76	Tooth agenesis	28933972 28933971 28933373
anodontia	k000	100	oligodontia-colorectal cancer syndrome	219097	100	Oligodontia-colorectal cancer syndrome	9913621 8081536

Continuation of Table

ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
anonychia	q843	82	anorchia, familial	198699	70	Familial colorectal cancer	10515446 2020383 4705609 4705610 10071425 464002 6594646 12518091 11950612 4705617 6891448 11241183 2439591 4705486 369952 10073398 2546116 4705624 28578275 11241184 9647583 7723423 6867243 12659119 2464806 74953290 35414976 62364016 11241185 2439589 392179 7707339 467033 396321 401908 62364017 35130225 2289485 2289484 2431242 2431238 66964065 1734243 2431514 2464803 517947 518013 2251913 529076 2952615 1613148 2545158 2546106 454886 390092 2707761 17164132 2545165 2545164 563556 2545163 6885768 2545160 2546107 2545159 2546110 544243 548710 569940 11960216 2909958 2909786 2546111 918397 458967 62626346 6899169 411356 1966477 1966476 866006 1804197 41116 448475 386830 448162
anterior soft tissue impingement	m2681	63	t-pa variant	85245	72	APOE2 VARIANT	429358 28931578
antiphospholipid syndrome	d6861	68	iscomatrix	159011	70	Pilomatrixoma	28931588
aphonia	r491	67	paridiformoside	60727	63	Hydatidiform mole	269938 269939 269940 269955
apraxia	r482	93	alzheimer disease, familial, 3, with spastic paraparesis and apraxia	219380	100	Alzheimer disease, familial, 3, with spastic paraparesis and apraxia	1800054 1800054 3218707 3218674 56123940 2235006 641252 1800056 3092857 3092842 1800058 1800889 1801673 1800061 1800061 28904921 55801750 55982963 55982963 17174393 17174393 1800558 28933383 28933383 28933383 28933382 63750599
arginosuccinic aciduria	e7222	71	lysinuric protein intolerance	218413	100	Lysinuric protein intolerance	121908678 72552272 146582474
arteriovenous fistula, acquired	i770	82	fabry disease	219125	100	Fabry disease	28935494 28935493 28935492 28935490 28935490 28935489 28935485 28935495 28935487 28935486 3027584
arteriovenous malformation, other site	q2739	68	glomangiomas, multiple	197892	64	Multiple fibrofolliculomas	41419545 8065832 142934950
asplenia (congenital)	q8901	74	transferrin/deficiency	199119	63	warfarin response - Efficacy	9923231
asplenia (congenital)	q8901	74	transferrin/deficiency	199119	63	warfarin response - Efficacy	
atelectasis	j9811	80	hypotrichosis-lymphedema-telangiectasia syndrome	219378	100	Hypotrichosis-lymphedema-telangiectasia syndrome	28936692 28936693
atopic neurodermatitis	l2081	87	dermatitis, atopic, 2	219999	100	Dermatitis, atopic, 2	61816761
atrial septal defect	q211	85	3c syndrome	196583	95	C syndrome	2274622 2274623 1410590 12568913 74315342 460897 696217 34911341 3816873 1042713 1042714 28936670 28936670 56298569 56298569 56208331 56208331 200783529 28933402 138909849 28999110 138924661 150038620 1800578 1800576
atrioventricular septal defect	q212	100	atrioventricular septal defect	218489	97	Atrioventricular septal defect 2	28942092 28942092 28942091 28942091
attention-deficit hyperactivity disorder, other type	f908	90	attention deficit disorder with hyperactivity	219436	94	Attention deficit hyperactivity disorder	61753973
autistic disorder	f840	100	adenylosuccinate lyase deficiency	199099	87	Argininosuccinate lyase deficiency	28941472
autoimmune hepatitis	k754	67	autoimmune polyendocrine syndrome, type 1	199132	85	Polyglandular autoimmune syndrome, type 1	34397615 72650677
autoimmune hepatitis	k754	67	autoimmune polyendocrine syndrome, type 1	199132	85	Polyglandular autoimmune syndrome, type 1	
autosomal recessive ocular albinism	e70311	63	heterotopia, periventricular, autosomal recessive	219357	100	Heterotopia, periventricular, autosomal recessive	3807153 62638194 1802492 28937880
bandemia	d72825	64	band 3 memphis	73589	100	Band 3 memphis	5036
barth syndrome	e7871	74	bardet-biedl syndrome	198820	100	Bardet-Biedl syndrome	35487251 121918327 121918327 121918327 506121 529208 28937575 185142838 55848325 35520756 113624356 113624356 148374859 148374859 148374859 2277598 121908177 121908177 4784677 4784677 4784677 28939088 28937875 28937875 16991547
bartter's syndrome	e2681	76	primary ciliary dyskinesia, 2	196548	96	Primary ciliary dyskinesia	142371860 142371860 142371860 144711161 121908171 72657316 35865357 201740530 200669099 201213030 201213030 59172778 139519641 57521499 11879596 11879596 143740376 143740376 143740376 151107532 151107532 151107532 138320978 138320978 138320978 41305223 62638654 62638631
basal cell carcinoma of anal skin	c44510	75	basal cell carcinoma, multiple	198601	72	Basal cell carcinoma, susceptibility to, 7	78378222
basophilia	d72824	61	scianna blood group antigens	144182	83	Antigen in Scianna blood group system	56047316
bence jones proteinuria	r803	65	cancer susceptibility candidate 3 protein, rat	146377	75	Prostate cancer susceptibility	35882952 8176058 138213197
benign neoplasm of bladder	d303	67	adenocarcinoma	197586	100	Adenocarcinoma	41341748
benign neoplasm of cranial nerves	d333	64	benign familial neonatal seizures	196705	97	Benign familial neonatal seizures 1	1801475 1801475 28939683 28939683
benign neoplasm of lip	d100	67	tag 72 (breast carcinoma)	52961	83	Ductal breast carcinoma	144848 144848 1799950 1799950
benign neoplasm of liver	d134	72	malignant mesenchymoma	196907	80	Malignant melanoma	16891982 1126809 861539 1805006 11547464 1110400

Continuation of Table

ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
benign neoplasm of prostate	d291	70	hereditary prostate cancer	198236	71	Hereditary cancer-predisposing syndrome	33927012 34916635 35882952 36053993 35352891 3219489 34126013 34612342 3219484 486907 74315364 74315364 1126497 56170584 17217716 63750614 63750002 35898375 61756462 63750124 63750255 63750716 63750843 1800151 63751455 63749832 34136999 4987188 4987188 4987189 63749849 17224367 63750086 63750820 63750330 63751207 63751207 63751207 3732183 61756466 61756467 63751236 63750280 61756468 63750875 63749932 63749932 63750032 63750790 63750398 63749811 63750810 63749854 63750368 63750684 63749983 63750027 63750797 41295182 63750664 1800932 63750143 63750143 3211299 1800935 1800937 63750955 1800938 63749980 63750878 61753793 55882234 63751405 41295268 61754783 63750897 63751005 63749874 63749973 41295270 63751121 3136334 63749886 63749886 63750832 63751419 56371757 63750138 63750138 63749895 63751450 63751321 63751321 34374438 2020912 63751408 63751017 63751113 63750258 63749999 63750617 3136351 3136351 3136351 63750753 63750753 63750753 2020910 63751327 63751328 63750949 63750370 63749942 63750119 63750119 63750882 63750836 63750767 41295278 721048 721048 28997576 2070094 1048108 5030826 5030807 5030829 5030809 5030834 5030817 5030824 35032294 1800734 41285097 63750648 63750656 63750952 61751642 4647220 63749924 63749887 2308317 1799977 35908749 63750642 63750642 63749896 63750796 63750268 63751049 63751049 63750710 63750447 63750430 63750749 63750791 63750365 34213726 63750540 63751145 63749909 41295284 63751247 63751214 35045067 63750109 63750899 63750610 1800146 55907433 63750014 63750292 63750242 63750809 63750217 63750217 35831931 142441643 74953290 62619935 62619935 1801155 866006 866006 34919187 2229994 2229995 72541816 28903085 28903088 1800562 17420802 63751422 63750451 1805324 63750490 63750250 1805318 63750947 63750770 63751211 63750055 63751300 63751028 1805320 1805323 63751228 1805321 12532895 143162541 10254120 10254120 63750123 11762213 13223756 34589476 41736 143153871 41341748 41341748 72552387 769420 34767364 1805794 61754966 61754795 61753720 200287925 1063045 3731249 6413464 34170727 11552822 36204594 4962081 35118262 9282835 1800860 9282834 1799939 1800863 17158558 10993994 10993994 10993994 607969 137852761 1800054 2234997 3218707 3218674 56123940 2235006 641252 1800056 3092857 3092842 3092856 1800058 1800889 1801516 1801673 1800061 1800061 28904921 55801750 55982963 17174393 17174393 17174393 1800558 34677591 11214077 4987046 28897705 766173 766173 11571640 28897706 144848 41293475 56403624 28897708 28897709 28897710 11571653 28897715 2227943 28897717 28897718 1799944 55638633 41293477 28897719 28897719 1801406 28897720 28897722 28897723 55969723 28897727 28897728 41293491 28897731 56087561 28897735 11571657 41293497 41293497 41293497 55875643 4987117 28897737 1799954 56191579 56191579 34309943 35029074 35029074 55953736 55953736 28897742 55977008 4986859 28897744 9534262 28897745 28897747 41293511 41293513 41293513 41293513 28897749 28897750 11571747 28897753 28897754 28897754 11571769 28897755 28897756 45580035 41293521 28897758 28897759 61757642 11571831 11571831 11571833 28897761 28897761 200355697 45517097 45517098 45517100 45517106 45505405 45517112 45473698 45442896 1800748 45517141 1800725 1800742 45478593 45517162 45484298 45517170 34012042 45517203 45517208 45517215 45517215 45517221 45517221 45517232 45517238 13337626 45517259 45517264 45517272 36078782 36078782 45517284 45478393 45487691 45517294 45483391 45486193 45517317 45517320 45517323 45438898 45474795 35986575 45517354 45517368 45517378 35534817 45475501 45517419 45517423 1800124 16260 33935154 2276330 35572355 35993958 35993958 35993958 55819519 28934574 28934574 28934576 28934576 11540652 11540652 28934573 1800372 28934578 28934578 55863639 1042522 1042522 1800371 1800370 4792311 41419545 17887014 28363284 4796033 41293465 45553935 41293461 28897696 28897696 55770810 55770810 8176258 1799967 1800726 8176219 1799966 1799966 28897693 56158747 28897692 28897691 28897691 55815649 1800744 41293455 41293455 1060915 28897690 28897689 55639854 28897686 28897686 55930959 55725337 62625307 28897685 16942 55909400 4986852 16941 1800704 56321129 56321129 56321129 799917 799917 41286300 28897684 1800709 28897683 56082113 28897682 62625306 16940 41286296 4986846 4986846 4986845 4986845 4986844 4986850 28897680 8176154 1800064 56039126 56012641 28897677 28897676 62625300 56128296 1799950 8176153 28897675 1799965 34545365 28897672 1800062 138213197 4986763 4988356 4986764 4986765 4988349 4988346 28903098 28903098 9913621 59912467 59912467 72552322 17879961 17879961 28909982
benign neoplasm of stomach	d131	74	carney triad	220351	100	Carney triad	142441643
benign neoplasm of stomach	d131	74	carney triad	220351	100	Carney triad	

Continuation of Table

ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
beta thalassemia	d561	100	delta-thalassemia	218427	100	delta Thalassemia	33985472 35949130 63750954 63751128 35703285 34690599 34690599 35328027 35328027 1609812 34451549 34451549 34451549 34937014 63751218 11549407 35348864 63750915 35004220 35004220 35004220 35724775 33915217 33915217 33915217 33915217 33915217 33915217 33971440 33971440 33971440 33960103 33960103 33960103 35477349 33950507 33950507 33986703 35662066 35662066 63750783 34856846 35699606 35699606 35497102 35497102 34889882 63750475 63750628 34305195 63750400 34598529 34598529 34598529 34500389 33944208 33944208 33944208 34999973 34999973 34999973 34883338 63751208 63751208 35324967 63750345 35887507 35654785 35152987 35518301 34975911
biotinidase deficiency	d81810	69	apnea, postanesthetic	198389	100	Postanesthetic apnea	2297950 34885143 13073139 28934601 35034250 13078881 28933389 1799807
bipolar ii disorder	f3181	91	bipolar affective disorder, early-onset	220794	78	Bipolar affective disorder, susceptibility to	121918300 6265
bone marrow transplant failure	t8602	75	anemia, aplastic	197668	100	Aplastic anemia	34094720 61754966 147948835 35947132
bone transplant status	z946	61	lymphomyosot	180778	71	Lymphoma, somatic	7732671 121908981
brown-sequard syndrome	g8381	74	cardiac conduction defect, nonspecific	221537	75	Cardiac conduction defect, susceptibility to	45489199 41261344 41261344 9858585 7626962 1805125 45553235 45553235 72549410 36210420 45471994 45471994 45620037 41276525 28933378 28933378 28933377 41314354 16924297 203462
budd-chiari syndrome	i820	100	budd-chiari syndrome	218567	61	Budd-Chiari syndrome, susceptibility to, somatic	6025 77375493 77375493
burkitt lymphoma, spleen	c8377	82	burkitt lymphoma	199276	100	Burkitt lymphoma	28933407
cafe au lait spots	l813	100	legius syndrome	207215	100	Legius syndrome	7180446 3751526
calculus of kidney	n200	81	pachyonychia congenita	198276	86	Pachyonychia congenita type 2	60627726 60944949 58181827 28928895 57424749 60723330 59349773 59349773 59977263 28933089 57674130 28933088 59151893 28928898
california encephalitis	a835	75	aicardi-goutieres syndrome 1	196824	100	Aicardi Goutieres syndrome 1	72556554
carcinoid syndrome	e340	73	retinitis pigmentosa	5537	100	Retinitis pigmentosa	62637004 61752909 61752878 61752871 61751281 61751407 61750152 61751263 61751386 61748550 41265017 28939720 62635654 62635656 62636275 62636291 146733615 200691042 199867882 28933395 28933394 28933993 29001637 121918579 137853907 34094720 61755817 61755816 61755786 201493928 1126809 61747071 28939088 73415876 62642057 62638651 62638644 62638637 62638634 62638632
carcinoma in situ of colon	d010	76	colon carcinoma kinase 4	96806	76	Carcinoma of colon	36053993 3219489 34612342 28936407 1799807 1566734 56079734 41419545 28937578 28358582
carcinoma in situ of endocervix	d060	65	carcinoma, neuroendocrine	197961	71	Endometrial carcinoma	36053993 34612342 6339 63751017 55907433 17158558 7080536 28756990 33935154
carcinoma in situ of trachea	d021	71	urachal cancer	197587	71	Colorectal cancer	63750124 63750255 34136999 63750875 3211299 63750955 63750878 41295268 63751005 63751234 63751450 34374438 2020912 41295278 34833812 63751049 63750365 10515446 2020383 4705609 4705610 10071425 464002 6594646 12518091 11950612 4705617 6891448 11241183 2439591 4705486 369952 10073398 2546116 4705624 28578275 11241184 9647583 7723423 6867243 12659119 2464806 74953290 35414976 62364016 11241185 2439589 392179 7707339 467033 396321 401908 62364017 35130225 2289485 2289484 2431242 2431238 66964065 1734243 2431514 2464803 517947 518013 2251913 529076 2952615 1613148 2545158 2546106 454886 390092 2707761 17164132 2545165 2545164 563556 2545163 6885768 2545160 2546107 2545159 2546110 544243 548710 569940 11960216 2909958 2909786 2546111 918397 458967 62626346 6899169 411356 1966477 1966476 1801155 866006 1804197 41116 448475 386830 448162 63750451 6964587 28756990 28756986 28937871 28937870 55770810 9913621 8081536 72552322 17879961
cardiac arrhythmia, unspecified	i499	73	cardiac arrhythmia, ankyrin-b-related	221126	100	Cardiac arrhythmia, ankyrin B-related	72544141
castleman disease	d47z2	74	cutis laxa with severe pulmonary, gastrointestinal, and urinary abnormalities	221641	100	Cutis laxa with severe pulmonary, gastrointestinal, and urinary abnormalities	12150220 1131620 1051303
cat-scratch disease	a281	65	charcot-marie-tooth disease, type 2e	198892	100	Charcot-Marie-Tooth disease type 2E	28940296 28940291 142271930 41313880 60864230 59885338 58105277 61217436 61444459 80338934 80338925 59443585 62636505 28928910 61491953
celiac disease	k900	80	abc disease	231418	74	Celiac disease 3	231775
central corneal opacity, left eye	h1712	72	neuromuscular disease, congenital, with uniform type 1 fiber	221246	100	Neuromuscular disease, congenital, with uniform type 1 fiber	63749869
cerebral amyloid angiopathy	i680	82	cerebral amyloid angiopathy, app-related	198850	100	Cerebral amyloid angiopathy, APP-related	63750921 63750921 63749810 63751039 63750671
cerebral atherosclerosis	i672	77	intimal medial thickness of internal carotid artery	218987	89	Intimal medial thickness of internal carotid artery, modifier of	3827760 1805192
cholecystitis, unspecified	k819	60	cholecystokinin 1 receptor, mouse	174047	82	CHOLECYSTOKININ A RECEPTOR POLYMORPHISM	52795588
chondrodysplasia punctata	q773	100	x-linked chondrodysplasia punctata 1	231556	86	Chondrodysplasia punctata 2 X-linked dominant	28935474 28935174
chronic atrial fibrillation	i482	83	atrial fibrillation, familial 1	199121	98	Atrial fibrillation, familial, 13	1805123 16969925 16969925
chronic graft-versus-host disease	d89811	74	granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type ii	220157	97	Chronic granulomatous disease, autosomal recessive cytochrome b-positive, type 2	13306575 13306575
chronic obstructive pulmonary disease, unspecified	j449	86	pulmonary disease, chronic obstructive	219571	80	Pulmonary disease, chronic obstructive, susceptibility to	1051740

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
chronic pulmonary embolism	i2782	82	schimke immunoosseous dysplasia	197719	100	Schimke immunoosseous dysplasia	119473033
citrullinemia	e7223	100	citrullinemia	197525	79	Citrullinemia type I	35269064
cleft hard palate	q351	83	cleft palate with ankyloglossia	219446	100	Cleft palate with ankyloglossia	57318642 59886214 61064130 58596362 59267781 28933081 28935177
combined immunodeficiency, unspecified	d819	84	severe combined immunodeficiency with sensitivity to ionizing radiation	198543	100	Severe combined immunodeficiency with sensitivity to ionizing radiation	1494558 1494555 121908156
congenital absence of ovary, bilateral	q5002	84	congenital bilateral absence of the vas deferens	197160	100	Congenital bilateral absence of the vas deferens	78655421 1800098 11971167
congenital agranulocytosis	d700	100	neutropenia, severe congenital, autosomal recessive 3	198545	100	Severe congenital neutropenia 3, autosomal recessive	74315322 74315322 74315322
congenital aneurysm of aorta	q2543	83	aortic aneurysm	220317	100	Aortic aneurysm	7579903 1800255 34833812 35766612 35766612 35766612 2228048 2291628 190450 32209 154001 363811 363835 140649 140630 140598 2228241 140587 55831697 2075511 16967494 2272554 4781689 2230267 2235491 1801181 2228298 5742905 234706
congenital bronchiectasis	q334	72	bronchiectasis	221577	100	Bronchiectasis	5742912 5742912 61759861 61759861 5736 5736 5738 5738 72654338
congenital bullous ichthyosiform erythroderma	q803	90	ichthyosiform erythroderma, congenital	197658	79	Bullous ichthyosiform erythroderma	28939077 58026994 60035576 60118264 58414354 58075662 57784225
congenital cataract	q120	100	cataract/congenital	196604	100	Congenital cataract	28928900 60864230 61214927 58672172 61282106 57920071 59981161 60890628 57830985 28931604 72551362 72551363 72551364 200190472 140207606 2231142 72552713 2231137 56141211 3735231 3735231 72549406 72549406 72549406 72549406 72549405 72549405 2070074 142090709
congenital central alveolar hypoventilation syndrome	g4735	91	congenital central hypoventilation syndrome	197358	88	Congenital central hypoventilation	3026785 8192466
congenital cirrhosis (of liver)	p7881	73	leber congenital amaurosis 4	220331	100	Leber congenital amaurosis 4	62636300 61752904 6339 121918165 147014855 57749775 61751266 17103671 28940314 28940315 62637016 62637015 62637014 62637012
congenital deformity of finger(s) and hand	q681	76	camptodactyly and knuckle pads	221688	63	Cataplexy and narcolepsy	545973 1551570 2305795
congenital diaphragmatic hernia	q790	78	spondylocostal dysostosis	198522	96	Spondylocostal dysostosis 2	71647808 71647806
congenital dilation of aorta	q2544	74	larsen syndrome, recessive type	198788	72	Insulin resistance syndrome, type A	60662302
congenital dyserythropoietic anemia	d644	100	majeed syndrome	198761	100	Majeed syndrome	41417446 34378160 28936671 28936672 34676691
congenital entropion	q102	69	insensitivity to pain, channelopathy-associated	220116	64	Thyroid-associated orbitopathy, susceptibility to	231775
congenital glaucoma	q150	100	friedreich ataxia	198954	94	Friedreich's ataxia	28936694 28936701 28936700 35703638 34595252 56214919
congenital hydronephrosis	q620	89	nephrotic syndrome	196966	100	Nephrotic syndrome	2274622 2274623 1410590 12568913 74315342 138909849 138909849 150038620 150038620
congenital hypertonia	p941	71	17,20-lyase deficiency, isolated	221193	100	Isolated 17,20-lyase deficiency	35887327 28940283 62269092 6472 28934586 61754278
congenital hypotonia	p942	68	charcot-marie-tooth disease	196572	100	Charcot-Marie-Tooth disease	28940296 28940291 142271930 121913615 41313880 60864230 60864230 59885338 58105277 61217436 28928902 57318642 61444459 6166 6165 80338934 80338925 28936670 59443585 59101996 58982919 62636505 28928910 28928910 61491953 28937906 28941470 28936370 121908866 199752932 119472026 9282774 28936682 80338732
congenital lactase deficiency	e730	72	left ventricular noncompaction, nonisolated	219601	81	Left ventricular noncompaction 7	28937595 28936670 41309764 201850378
congenital leukonychia	q844	77	autosomal recessive pachyonychia congenita	198981	82	Autosomal recessive congenital ichthyosis 1	726070 28940568 28940271 28940270 28940269 35312232 41295338
congenital methemoglobinemia	d740	72	hemoglobin contaldo	48689	100	HEMOGLOBIN CONTALDO	33950507 41341344 41417548 41412046 35329201
congenital mitral insufficiency	q233	76	adrenal insufficiency, congenital, with 46,xy sex reversal	220555	85	Adrenal insufficiency, congenital, with 46,XY sex reversal, partial or complete	72547508
congenital myopathies	g712	79	minicore myopathy with external ophthalmoplegia	219779	100	Minicore myopathy with external ophthalmoplegia	62636495 62636495 60798368 57496341 57496341 57639980 57639980 58898021 57955682 57955682 73991549 73991549 28936383 28937594 28937594 62541771 62541771 200783529 28933402 201754030 28999110 200563280
congenital night blindness	h5363	87	atrophia bulborum hereditaria	198770	100	Atrophia bulborum hereditaria	62636300 61752904 62636273 62636273 62636275 62636275 61755802 61755802 61755802 121918165 61750420 61751266 17103671 28940314 28940314 28940315 28940315 62637016 62637015 62637014 62637012 61749755 61749755 61750187 61750187 28933685
congenital non-neoplastic nevus	q825	76	giant pigmented hairy nevus	197885	100	Giant pigmented hairy nevus	11554290
congenital nonprogressive ataxia	g110	70	spinocerebellar ataxia, autosomal recessive 1	198294	100	Spinocerebellar ataxia autosomal recessive 1	29001665
congenital nystagmus	h5501	100	nystagmus 2, congenital, autosomal dominant	198774	79	Congenital myotonia, autosomal dominant form	55960271
congenital ptosis	q100	80	death, sudden	197362	67	Sudden unexplained death	45471994 34094720 34094720 36210421 139617644 28935490
congenital renal failure	p960	71	seizures, familial febrile, 3a	221706	100	Febrile seizures, familial, 3a	3812718
congenital rubella syndrome	p350	83	horner syndrome, congenital	219278	83	Congenital long QT syndrome	2274622 2274623 1410590 12568913 74315342 45609733 72549410 45546039 72544141 28928904 28928904 28928904 731506 179489 179489 17215479 12720449 17221854 34516117 17857111 28933384
congenital viral disease, unspecified	p359	72	myopathy, myosin storage	219329	100	Myosin storage myopathy	28933098

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
congenital viral hepatitis	p353	68	px protein, hepatitis b virus	62339	70	Hepatitis b virus, susceptibility to	2229207 2834167
conjoined twins	q894	67	pannexin 2, rat	148763	63	Anorexia nervosa 2	6265
conn's syndrome	e2601	86	cockayne syndrome	218307	100	Cockayne syndrome	28928874 1800124
copper deficiency	e610	100	seizures	196706	100	Seizures	74315376 2298771 6432860 3812718 6746030 142881576 12530380 6476 6472 7755898 72554626 61757294 61757294 28931609 138326449 138326449 140621530 140621530 2303790 28941476 28999113 1801475 28939683 28935479
cori disease	e7403	77	beta-ureidopropionase deficiency	218642	96	Deficiency of beta-ureidopropionase	1056836 4444903 61755793 61755792 33950507 33930165 35333334 34002892 200646278 138081800 34035085
coronary artery dissection	i2542	86	coronary artery disease	218862	79	Coronary artery disease, modifier of	1801278 3732378 3732379 662 1024611 1024611
cracked nipple associated with lactation	o9213	64	lung cancer marker lcap	107601	71	Small cell lung cancer	11554290 11554290
craniofacial dysostosis	q751	82	craniofrontonasal dysplasia	197572	100	Craniofrontonasal dysplasia	28936071
craniosynostosis	q750	94	baller-gerold syndrome	197858	100	Baller-Gerold syndrome	17217716 4987188 63750379 63751207 3732183 2303428 63749811 41295182 63749873 63750897 63751005 63750706 1799977 63750796 63751049 63750710 63750710 63750447 63750365 34213726 63750059 63751247 63751247 63750899 63750899 1800146 63750217 63750217 17420802 63750250 1805318 1805321 10254120 63750123 34666647
cr(e)st syndrome	m341	76	becker nevus syndrome	220299	80	Epidermal nevus syndrome	11554290
crigler-najjar syndrome	e805	100	crigler najjar syndrome, type 2	197361	97	Crigler Najjar syndrome, type 1	34983651 72551341 62625011 72551349 72551351
cutaneous amebiasis	a067	74	neurocutaneous melanosis	198363	100	Neurocutaneous melanosis	11554290
cutaneous anthrax	a220	71	9-amtm	103545	71	Autism 9	1858830
cyclophoria	h5054	61	tpicc cpd	194063	74	G6PD TEPIC	1050829 1050828
cyclosporiasis	a074	85	cyclosporine metabolite m17	57047	67	cyclosporine response - Dosage, Metabolism/PK	776746
cysticercosis of central nervous system	b690	84	leukoencephalopathies	6947	90	Leukoencephalopathy	142433332 201330912 28939079 201258663 28933698 28937321 11568188
cystinuria	e7201	100	cystinuria	193494	100	Cystinuria	28934891 121964962 5742905
day blindness	h5311	82	blindness	197842	100	Blindness	62638625 62638624 62638214 62638208 62638202 62637021 62637037
deficiency of vitamin e	e560	75	heparin cofactor ii deficiency	218504	100	Heparin cofactor II deficiency	35916840 5907
deletion of short arm of chromosome 4	q933	82	8p deletion syndrome (partial)	198748	69	Ovarian hyperstimulation syndrome	6166 28928871 28928870
delusional disorders	f22	64	bleeding disorder, east texas type	219986	66	Platelet-type bleeding disorder 13, susceptibility to	34377097
demyelinating disease of central nervous system, unspecified	g379	77	megalencephalic leukoencephalopathy with subcortical cysts	197296	98	Megalencephalic leukoencephalopathy with subcortical cysts 1	72466451 11568188
dental restoration status	z98811	65	mental retardation, x-linked, syndromic 13	221036	100	Mental retardation, X-linked, syndromic 13	28935479 28933691 61753980 3027927 61753973 61753971 61753968 61753965 61753016 61751449 61751444 28935468 61751439 61751362 61750240 61750240 61749721 61749715 61749720 61748426 61748420 28934906 28934906 61748408 28934908 61748392 28934904
dermatographic urticaria	l503	60	dermorphin receptors	59428	68	morphine response - Dosage	2952768 1045642
deuteranomaly	h5353	73	pi 39	16295	80	PI M3	1800571 1303
developmental disorders of jaws	m270	69	acromesomelic dysplasia, maroteaux type	196873	100	Acromesomelic dysplasia Maroteaux type	28931581 28931582 28929479
diabetes insipidus	e232	100	diabetes insipidus	197131	100	Diabetes insipidus	71524377 28937890 28931580 28934878
diastrophic dysplasia	q775	100	diastrophic dysplasia	197323	100	Diastrophic dysplasia	104893915 3776070
di george's syndrome	d821	92	digeorge syndrome-velocardiofacial syndrome complex 2	218700	75	Conotruncal anomaly face syndrome/velocardiofacial syndrome	28934568 28934568 35766612 28939675
dilated cardiomyopathy	i420	100	idiopathic dilation cardiomyopathy	197419	77	Cardiomyopathy, idiopathic dilated, mitochondrial	57077886 57077886 28928903 58922911 28928900 28928900 59270054 59270054 41313880 59026483 59026483 61195471 61195471 61195471 28933092 28933092 28933092 61295588 61295588 60682848 60682848 60682848 56816490 56816490 56771886 56771886 56851164 56851164 59301204 59301204 61672878 61672878 534807 534807 56984562 56984562 56984562 56984562 56984562 61444459 61444459 60890628 60890628 63750197 63750197 6716782 6716782 201273719 62636495 62636495 62636491 62636492 62636492 73991549 63750743 45546039 45546039 45546039 45620037 45620037 34768413 147622517 147622517 1417635 1417635 34580776 34580776 34580776 2228387 2228387 2228387 35404804 35404804 61001398 61001398 193922674 1046116 201754030 201754030 45464193 45516091 45516091 45614536 45614536 45578741 45578741 199896820 199896820 3134560
disorders of ketone metabolism	e7132	83	disorder of isoleucine metabolism	197967	69	Debrisoquine, ultrarapid metabolism of	1799971 4986893 4244285 1135840 16947
disseminated superficial actinic porokeratosis (dsap)	l565	93	porokeratosis, disseminated superficial actinic 1	197471	100	Porokeratosis, disseminated superficial actinic 1	104895362
drug induced constipation	k5903	62	chlorproguanil - dapsone	182356	63	chlorproguanil and dapsone response - Toxicity/ADR	1801155 339097 339097 12777823 12777823 4244285 1799853 1799853 1799853 7900194 7900194 4917639 4917639 1057910 1057910 1057910 1057910 28371686 28371686 7294 7294 2359612 2359612 8050894 8050894 9934438 9934438 17708472 17708472 2884737 2884737 9923231 9923231 9923231 9923231 7196161 7196161 1876828 2108622 2108622 1801272 1050828
drug-induced tremor	g251	61	phenytoin receptors	37196	81	Phenytoin response	3812718 4244285 1057910
dysphonia	r490	82	dystonia 13, torsion	219392	73	Dystonia 1, torsion, modifier of	1801968
dysthymic disorder	f341	83	dystonia, dopa-responsive	198905	100	Dystonia, dopa-responsive	3812718 1045642 1801968 1801968 1800499 41298442 41298442 41298442

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
ectodermal dysplasia (anhidrotic)	q824	78	oculoectodermal syndrome	219144	100	Oculoectodermal syndrome	35136134 35136134 28937872 28933100 200446614
ehlers-danlos syndrome	q796	100	ehlers-danlos syndrome type 1	197344	95	Ehlers-Danlos syndrome, type 7A	121913552 121913552 6434312 6434312 76148000 76148000 4128539 4128539 28937869 28937869 137853146 137853146 72656354 72656354 66820119 66820119 72656357 72656357 67162110 67162110 72659324 72659343 72659343 61735045 61735045 72645347 72645347 72667022 72667022
elevated c-reactive protein (crp)	r7982	61	adrenergic receptor kinase, beta 2, mouse	180395	56	selective beta-2-adrenoreceptor agonists response - Efficacy	7793837
elevated c-reactive protein (crp)	r7982	61	adrenergic receptor kinase, beta 2, mouse	180395	56	selective beta-2-adrenoreceptor agonists response - Efficacy	
enchondromatosis	q784	93	chondromatosis	218536	79	dyschromatosis	28936681 28936680
endocarditis in systemic lupus erythematosus	m3211	80	neonatal systemic lupus erythematosus	197524	77	Systemic lupus erythematosus, susceptibility to	2476601 5744168 7574865 231775 72556554 10516487 2004640 2070197 10954213 1053874
endometriosis, unspecified	n809	61	erythrocyte p57 proteinase	90736	63	Polyagglutinable erythrocyte syndrome	7080536 17261572
endothelial corneal dystrophy	h1851	97	maumenee corneal dystrophy	197560	79	Meesman's corneal dystrophy	57872071 60410063 58162394 58918655 57218384 57218384 58410481
enteroviral meningitis	a870	62	retinitis pigmentosa 1	199221	100	Retinitis pigmentosa 1	62637004 62637004 61752909 61752909 61752878 61752878 61752871 61752871 61751407 61751407 61750152 61750152 61751263 61751263 61751386 61751386 61748550 61748550 41265017 28939720 28939720 62635654 62635654 62635656 62635656 62636275 62636275 62636291 62636291 200691042 200691042 61755816 61755786 61747071 62642057 62638651 62638644 62638637 62638634 62638632
eosinophilia	d721	100	ofuji's disease	197134	76	Pick's disease	2301612 28929478 63751068 28934578 63750129 63750635
eosinophilic esophagitis	k200	80	limb-girdle muscular dystrophy type 2a	197088	78	Limb-girdle muscular dystrophy-dystroglycanopathy, type C5	58596362 57496341 73991549 28933693 28937905 28937900 28937903
epidermolysis bullosa dystrophica	q812	100	epidermolysis bullosa, pretibial	196729	70	Epidermolysis bullosa herpetiformis, Dowling-Meara	121912854 121912854 61348633 60715293 58330629 60399023 60171927 28928893
epidermolysis bullosa simplex	q810	100	epidermolysis bullosa simplex, ogna type	197140	75	Epidermolysis bullosa simplex, Cockayne-Touraine type	57348201 57599352 58072617 60586163 58163069 59115483 59115483 58058996 59629244 61371557 60725382 57358989 57358989
eructation	r142	63	epidermal ichthyosis bullosa of siemens protein, human	89919	71	Ichthyosis bullosa of Siemens	56829062 57510142
erythrasma	l081	64	erythrocytes	20211	85	Erythrocytosis	5030834 5030824 28940301 28940300 77375493
essential (primary) hypertension	i10	85	hypertension, essential	218286	100	Essential hypertension	699 5186 13333226
essential tremor	g250	100	essential tremor	197648	100	Essential tremor	6280
essential tremor	g250	100	essential tremor	197648	100	Essential tremor	

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
evans syndrome	d6941	77	angelman syndrome	193478	100	Angelman syndrome	56170584 17217716 63750589 63750614 63749907 63750966 63750777 63751246 63751056 63751260 63750334 63750644 63750087 1800150 63751158 63751429 63750002 63750230 63750318 63751173 35898375 61756462 63750408 63750401 63750124 63751226 63751426 63750624 63750255 63750716 63750843 63750843 63750902 63750933 63750069 63751291 63750551 63750037 63750088 63750821 1800151 63751326 63750682 63750786 63750327 63751110 63751136 63749897 63750488 63750690 63751455 63750881 63750881 63749832 63750058 34136999 63751159 63750091 63750381 63750701 63750276 63751115 63750934 63750885 63750046 63750640 63750640 4987188 4987188 4987189 63750630 4952887 13425206 63751219 63750267 63750039 63750496 63749849 17224367 63750302 63750611 63750611 63751169 63749850 63751271 63751192 63750813 63750813 63751142 63750379 63750379 63750132 63750086 17036577 3771281 63750820 63750330 63750330 63750224 63751207 63751207 63751207 63750094 63750738 63750845 63750104 63750029 63750675 63750662 63750597 3732183 61756466 63750633 55778204 61756467 17036614 63751236 63750047 63750665 63750493 63749982 63750312 63750806 63750508 63750280 63750626 61756468 63750960 63750875 63749929 2303428 2303428 63749932 63750032 63749993 63750790 63749878 63750232 63751432 63750961 63750398 63749811 63750108 63751453 63750545 63750810 63750722 63750794 63749802 4608577 63751119 63749854 63749913 63750368 63750346 63751143 63750896 63750684 63750618 63750149 63750233 63750803 63750803 63749983 34986638 63750008 63750027 63749975 63750857 63750797 63750797 63749830 63749830 63750291 63751093 2042649 41295182 6544991 3136228 41294988 63750664 3136245 3136247 1800932 63750143 3211299 63749873 1800935 3136329 1800936 1800937 1800937 1800937 63750955 1800938 63750996 63749980 63749980 63750552 63750878 61753793 55882234 63750065 63751452 63751405 63750741 41295268 61754783 63750897 63751005 63749874 63751312 63750595 63749893 63749973 41295270 63750564 63751121 3136334 63750462 63749886 63751167 63749857 63750358 63750832 63750832 63751419 63750304 35552856 63751442 56371757 63750647 63750138 63750725 63749895 63749895 63749899 63750637 63751450 63751321 63750389 34374438 63750357 2020912 2020912 63751408 2020914 63749889 63750904 63751017 63750056 63750940 63751113 63750111 63750942 63750258 63749938 63750563 63750287 63749999 3136337 63749821 63750617 3136351 3136351 3136351 3136351 63750753 63750753 2020910 63750377 63751427 63751327 63751328 63750731 63750949 63750370 63749942 63750119 63750119 63750882 63751058 63750554 63750554 63750262 63750139 63751319 63750836 63750767 41295278 35032294 1800734 41285097 56198082 56198082 63750745 63749816 63751131 63749804 63750648 63750706 63750706 63749827 63750656 2020872 63750952 61751642 4647220 63749803 63750645 63750658 63750542 63749818 63749820 63750638 63749924 63751653 63749887 2308317 2308317 1799977 1799977 35908749 63750642 63750962 63749896 63751259 63749926 63750796 63750796 63750286 63750268 63751049 63750710 63750533 63749864 63750715 63749965 63750447 63750430 63750430 63750749 63751015 63751153 63751179 63750293 63750791 63750316 63750443 63751414 63750748 63750365 34213726 63749981 63750071 63750540 63750616 63750314 63750956 63751145 63751435 63749909 3774332 63750059 63750059 63750035 63750718 63751240 41295284 63750150 63751247 63751415 63750152 63751214 63750846 63751639 63750240 35045067 35045067 63750109 63750899 63750610 63751301 63751202 63751202 1800146 63750726 55907433 63751200 2241031 63750282 63750014 63750740 63750663 63750242 63750242 63750809 63750809 63750217 63750864 63751652 63750603 63750603 35831931 63750971 63751075 63749875 3822471 1669445 62654864 28931602 17420802 63751422 63750451 1805324 63750490 63750250 1805318 63750947 63750770 63751211 63750055 63750236 63751300 1805320 1805323 1805323 1805321 63750106 63750049 2286680 2345060 12532895 12532895 143162541 143162541 10254120 63750123 2234584 17782839 28756987 28756986 1801376 28989187 28989182 28989185 80338701 28936415 28936683 28933384 28933384 61753251 61753251 3027927 28935468 61749720 28934906 61748396 28934904 61754453
exocrine pancreatic insufficiency	k8681	100	exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis	221270	100	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis	119455950
fabry (-anderson) disease	e7521	74	fabry disease, cardiac variant	221181	100	Fabry disease, cardiac variant	12713559 6413458 72653077 12713844 1800206 28935485
failure to thrive (child)	r6251	85	failure to thrive	196802	73	Heart failure	61767072 1801253
familial erythrocytosis	d750	100	familial erythrocytosis, 1	197904	100	Familial erythrocytosis, 1	5030834 5030824 28940301 28940300 63750560 63750567 77375493
familial hypercholesterolemia	e7801	71	hyperlipoproteinemia, type 2 a	199322	86	Familial type 3 hyperlipoproteinemia	11583680 2483205 505151 3737787 3737787 2073658 2073658 12713559 5742904 533617 1801699 12713844 12691202 137852987 6180 268 28937579 2230806 2230806 1800853 104895097 61732874 28940579 28940578 28940578 61752717 61752717 28940580 28940580 11466045 104895083 11466023 3743930 17249141 6511720 2228671 2228671 2228671 72658858 72658860 72658860 11669576 13306498 28942078 28942078 28942078 28942079 28942079 5930 4508523 17248882 688 5926 5925 2569548 2569548 72658867 2569538 5928 5928 429358 769455 7412

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
familial hypophosphatemia	e8331	83	vitamin d-dependent rickets, type 1a	218414	99	Vitamin D-dependent rickets, type 1	12713559 6413458 72653077 12713844 61495246 28934607 28934605 28934604
fat necrosis of breast	n641	65	tumor necrosis factor-alpha, cngrc fusion protein, mouse	140229	67	Tumor necrosis factor alpha (TNF-alpha) inhibitors response - Efficacy	1800629
fear of blood	f40230	62	d antigen blood group	34714	91	DIEGO BLOOD GROUP ANTIGEN	2285644
fecal impaction	k5641	62	sanofi brand of oxaliplatin	37744	61	oxaliplatin response - Efficacy, Toxicity/ADR	25487
fistula of gallbladder	k823	71	gallbladder disease 4	221088	100	Gallbladder disease 4	11887534
food protein-induced enterocolitis syndrome	k5221	60	von hippel-lindau syndrome protein, human	162012	79	Von Hippel-Lindau syndrome	5030826 5030826 5030826 5030804 5030804 5030804 5030807 5030829 5030809 5030810 5030834 5030817 5030817 5030817 5030819 5030824
frontal encephalocele	q010	76	knobloch syndrome	198207	94	Knobloch syndrome 1	137853108 199821258 12483377
frotteurism	f6581	63	testis and spermatogenesis related 4 protein, human	147050	57	anthracyclines and related substances response - Toxicity/ADR	11554290 763110 17244841 909253 33980500 10455872 324981 3793784 2234767 11200638 671 16969968 1051730 1532624 2232228 1799990 1056892
fusion of spine, thoracolumbar region	m4325	60	butyrylcholinesterase	10611	82	Butyrylcholinesterase activity	1803274
galactorrhea	o926	70	bile acid malabsorption, primary	221599	100	Bile acid malabsorption, primary	72547505
galactosemia	e7421	70	glaucoma	196673	100	Glaucoma	28936694 28936701 9282671 28936700 35703638 34595252 76917243 1800546 28939688
gastrointestinal anthrax	a222	71	familial intestinal polyatresia syndrome	218311	74	Familial multiple polyposis syndrome	1801155 2229995
gastrointestinal stromal tumor of rectum	c49a5	85	gastrointestinal stromal tumors	198477	98	Gastrointestinal stromal tumor	1801131 1801133 33927012 34916635 1805087 1801394 1532268 162036 2071010 34677591 34677591 11214077 11214077 11540652 28934573 1051266
gastroparesis	k3184	67	deptropine methobromide	30643	62	Torsades de pointes	36210420 36210421 36210419
gastroschisis	q793	69	foveal retinoschisis	199223	81	Juvenile retinoschisis	2282440 2491132 61753174 61752068 61752067 61752063
gaucher disease	e7522	100	gaucher disease, norbottnian type	220180	66	Subacute neuronopathic Gaucher's disease	80356772 421016 421016 421016 421016 421016 1064651 1064651 1064651 1064651 1064651 1064651 2230288 1064644 364897 364897 364897 364897
generalized abdominal rigidity	r1937	64	berardinelli-seip congenital lipodystrophy, type 4, with muscular dystrophy	221589	71	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A5	28940869 28941782 200056620 104894093 28937900
generalized atherosclerosis	i7091	76	cutis laxa	193492	100	Cutis laxa	1802492 1131620 1051303
generalized edema	r601	76	epilepsy, idiopathic generalized	218416	97	Epilepsy, idiopathic generalized 6	2298771 2298771 6432860 6432860 6746030 6746030 6746030 60734921 58173258
generalized hyperhidrosis	r61	73	lentiginosis, inherited patterned	225934	64	Leanness, inherited	5030980
genetic susceptibility to other disease	z1589	70	tumor susceptibility gene 101 protein, rat	98894	73	Prostate cancer, susceptibility to	486907 699 1143627 6280 4917 4918 9264942 7076156 2279744 671 16260 1800566 1805127 17879961
gilbert syndrome	e804	84	meckel syndrome	197288	77	Melnick-Needles syndrome	137853108 199821258 62640570 62640570 28935473 28935472
glanders	a240	67	11-autms	6307	71	Autism 10	52815063 7794745 7794745 2710102 2710102 1861972 1861972 1861973 1861973
glare sensitivity	h5371	76	coumarin sensitivity	221328	67	Acute alcohol sensitivity	671
glutaric aciduria type ii	e71313	90	glutaric acidemia type 1	197896	92	Glutaric aciduria, type 1	11085825 121434369
glycosuria	r81	77	glycosuria, renal	221440	73	Familial renal glucosuria	61742739
guttate psoriasis	l404	62	glutathione peroxidase	635	77	GLUTATHIONE PEROXIDASE POLYMORPHISM	11209026 1050450
hemoglobin e-beta thalassemia	d565	71	hemoglobin beta-subunit, thr(87)-	135272	66	Fetal hemoglobin quantitative trait locus 1	11886868 4895441 35378915 35321913 35983258 35710727 35617911
hemoglobinuria	r823	75	hb sassari	59247	73	G6PD SASSARI	11886868 11556045 4895441 2070075 2070074 35117167 33935780 34980264 34407387 34407387 35825479 34095019 34188626 35834416 34049764 35485099 34945623 35871407 34022507 33972927 33952147 33952147 33952147 35693898 35914488 35960772 35960772 35286210 35452098 34362537 34151786 33991472 33991472 33935983 33935983 35094013 34037627 34037627 35303218 35262412 41417446 34378160 34831026 35973315 35857380 35857380 35685286 33950507 63749918 35474880 35474880 35890959 35203747 35203747 33930165 33930165 33930165 33971270 34420481 34289459 34460332 35395083 34012192 34313675 34313675 35849660 36049074 34747494 34432567 34432567 36006195 35378915 35378915 35321913 35983258 35710727 35617911 34817956 34410516 34890875 41461652 41461652 63750122 41510746 34440919 41484451 41484451 41331747 63750073 41417548 41479844 41479844 36030576 28928884 35329201 28928881 35974739 35166834 35723200 35723200 5030868
hemolytic-uremic syndrome	d593	100	hemolytic-uremic syndrome	193502	82	Atypical hemolytic-uremic syndrome 6	460897 138924661 138924661 1800578 1800576
hemophagocytic lymphohistiocytosis	d761	100	griscelli syndrome type 2	198288	100	Griscelli syndrome type 2	28933376 28933374 28933375 35947132 35947132 28938176
hemoptysis	r042	60	dendritic cell and monocyte chemokine-like protein, human	171571	57	Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency	45598538
hepatoblastoma	c222	79	retinoblastoma	220940	79	Hepatoblastoma	28931588 3092902 3092891 4151539 28934573
hereditary erythropoietic porphyria	e800	81	protoporphyria, erythropoietic	221465	100	Erythropoietic protoporphyria	28941774 2272783 3848519
hereditary factor viii deficiency	d66	77	intrinsic factor deficiency	218654	68	Hereditary factor VIII deficiency disease	6025 35211634 35211634 1800297 28933681 28933679 28933676 28933675 28937289 28937287 28933673 28937285 28937282 28937272 28933671 28933670 28933668 28933668 28933669
hereditary hemochromatosis	e83110	70	symmetric dyschromatosis of the extremities	196936	93	Symmetrical dyschromatosis of extremities	28936681 28936680

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
hereditary hemorrhagic telangiectasia	i780	100	telangiectasia, hereditary hemorrhagic, type 2	198146	100	Hereditary hemorrhagic telangiectasia type 2	28936688 28936401
hereditary hypogammaglobulinemia	d800	72	agammaglobulinemia, autosomal recessive	198950	97	Agammaglobulinemia 2, autosomal recessive	34983651 34983651 4148323 4148323 35003977 57307513 34946978 36076514 1064422
hereditary motor and sensory neuropathy	g600	100	dysautonomia, familial	218827	100	Familial dysautonomia	28940296 28940291 6746030 80338925 80338925 28940281 111033171
hereditary optic atrophy	h4722	75	optic atrophy, x-linked	198133	79	Leber's optic atrophy	121918244 28939082 80338732 1599988 28359178
hereditary sideroblastic anemia	d640	100	anemia, hereditary sideroblastic	197834	100	Hereditary sideroblastic anemia	28935484
hereditary spastic paraplegia	g114	100	optic atrophy	196581	100	Optic atrophy	28939082 71524377 28937890 146262009 146262009 121908613 121908613 200737038 28941778 2234584 17454674 200440467 200440467 28936671 28936672 28939094 28939094 141848292 141848292 141848292 36014111 36014111 36014111 3759871 3759871 3759871 61755320 61755320 72547552 72547552 2301600 2301600 2301600 2233695 2233695 1599988 28359178
hereditary spherocytosis	d580	77	alport syndrome, recessive type	197682	81	Alport syndrome, X-linked recessive	201118996 28933406 28931585 28931584 28931583 45562031 1006269
hermansky-pudlak syndrome	e70331	96	hermansky pudlak syndrome 2	198643	96	Hermansky-Pudlak syndrome 4	692003 72551314 72551315 72551316 72551317 72551318 72551319 59428328 72551320 41272687 72551322 72551322 201227603 201227603 2296436 2296436 11592273 11592273 61884288 61884288 119471023 119471023
hirschsprung's disease	q431	95	waardenburg syndrome, type 4c	221614	100	Waardenburg syndrome type 4C	3026906 3918290 28939716 28936670 2435357 1800858 17158558 5352 45484298 11570255 11570351 73415876
hirsutism	l680	100	hirsutism skeletal dysplasia mental retardation syndrome	197783	72	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	201213306
histidinemia	e7041	79	histidine kinase	11379	86	Increased histidine	141634423 34457757
homocystinuria	e7211	100	homocystinuria, pyridoxine-responsive	220149	100	Homocystinuria, pyridoxine-responsive	1801131 1801133 2952768 28934891 121964962 5742905
horner's syndrome	g902	82	orbeli syndrome	196721	87	Gorlin syndrome	28933408 62637631
horner's syndrome	g902	82	orbeli syndrome	196721	87	Gorlin syndrome	
hydromyelia	q064	67	ornithine carbamoyltransferase	39276	85	Ornithine carbamoyltransferase deficiency	72554308 72554312 67939655 67486158 72554331 72554332 72554338 72554345 67960011 68026851 66741318 72556267 66626662 67367843 72558412 72558423 67120076 72558431 72558454
hypercalcemia	e8352	100	hypercalcemia, idiopathic, of infancy	218362	100	Idiopathic hypercalcemia of infancy	201304511 28934583 28934583 28934582 6068812
hypercarotinemia	e671	85	sarcosine dehydrogenase/deficiency	198230	91	Proline dehydrogenase deficiency	80338897 80338898 80338899 11555096 121965075 2904552
hypercementosis	k034	74	cortical hyperostosis with syndactyly	198483	76	Infantile cortical hyperostosis	72653170
hypercementosis	k034	74	cortical hyperostosis with syndactyly	198483	76	Infantile cortical hyperostosis	
hyperkalemia	e875	82	hyperlexia	220139	80	Hyperekplexia 3	121908493 121908493
hyperprolactinemia	e221	77	hyperlipoproteinemias	218898	98	Hyperlipoproteinemia	11591147 28942111 28362270 12713559 144467873 6413458 72653077 12713844 429358 769455 7412
hypersensitivity angiitis	m310	78	purine-pyrimidine metabolism, inborn errors	197620	65	heroin response - Metabolism/PK	1142345 1800584 1800460 1800462 1061235 1799971
hypertelorism	q752	100	roberts-sc phocomelia syndrome	196895	100	Roberts-SC phocomelia syndrome	34983651 4148323 72551349 4732748
hyperventilation	r064	79	dermatopathia pigmentosa reticularis	196637	100	Dermatopathia pigmentosa reticularis	72466487 72466486 67586389 67586389 8192466 60831116
hypocalcemia	e8351	83	hyperinsulinemic hypoglycemia, exercise-induced	199228	70	Persistent hyperinsulinemic hypoglycemia of infancy	62269092 72559713 28936371 72559716 28938469 28936370 151344623 72559722 72559723 4148619
hypopituitarism	e230	100	septo-optic dysplasia	193529	82	Septo-optic dysplasia sequence	28936702
hypoplastic left heart syndrome	q234	71	prostate cancer, hereditary, 12	221506	98	Prostate cancer, hereditary, 2	33927012 33927012 34916635 721048 34231037 34231037 142441643 28936670 34255532 34677591 11214077 4792311
hyposplenism	d730	67	hypogonadotropic hypogonadism 7 with or without anosmia	218467	90	Hypogonadotropic hypogonadism 8 without anosmia	28933074 144292455 28939719
ichthyosis vulgaris	q800	100	ichthyosis vulgaris	197232	100	Ichthyosis vulgaris	61816761
idiopathic aplastic anemia	d613	73	radial-ulnar hypoplasia with bone marrow failure and-or leukemia	197825	59	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1	34094720
immunodeficiency, unspecified	d849	73	leukocyte adhesion deficiency type 1	197081	100	Leukocyte adhesion deficiency type 1	2280965
imperforate hymen	q523	61	integrin, alpha m (complement component 3 receptor 3 subunit), human	172291	70	COMPLEMENT COMPONENT 8, ALPHA SUBUNIT, A/B POLYMORPHISM	652785
inflammatory disorders of seminal vesicle	n490	64	androgen-responsive protein f, rat	128814	62	efavirenz response - Dosage	2952768 1045642 3745274
iniencephaly	q002	80	lissencephaly	197202	93	Lissencephaly 1	6628 6628
intestinal myiasis	b8782	74	carcinoid tumors, intestinal	218497	87	Carcinoid tumor of intestine	34677591 11214077
intestine transplant infection	t86852	65	responsive-to-antagonist1 protein, arabidopsis	155895	59	alfentanil response - Metabolism/PK	1799971

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
iron deficiency	e611	74	enterokinase deficiency	218394	100	Enterokinase deficiency	1051740 63751247 63750899 28933395 28933394 28933993 29001637 121918579 121918579 137853907 6189 1799971 63751422 63750106 1800450 5030737 1800497 1799970 5030861 5030860 5030859 5030858 5030857 62507322 62509015 62516142 5030856 62642937 62642941 62516096 5030654 62507344 5030855 62508646 62516092 62514959 62508578 62642936 62642935 62642934 62514956 5030853 62642933 62642939 62516146 5030852 62508698 62514953 62514950 62508752 5030849 5030850 5030850 62642930 5030847 62508588 5030846 62507348 5030845 62514936 62514934 62514928 62514927 62507336 5030843 62507321 62517166 76296470 62508727 62514902 75193786 62514898 5030841 62642926 62514895 62514895 62642906 62514893 56127440 72553883 28730837 56378716 142150953 77200626 138221037
isolated proteinuria	r800	62	gpi-linked proteins	11463	61	G6PD PETRICH-LIKE	28940885 28940884 28940883 5743618 1229984 1800629 7779029 1517114 4986893 4244285 1800497 28989187 28989182 28989185 1051730 147210405 1042522 1042522 1876828 3745274 25487 25487 3212986 3892097 72554664
isosporiasis	a073	64	isoniazid	22886	71	Insomnia	1799990 28933385
isovaleric acidemia	e71110	100	isovaleryl-coa dehydrogenase deficiency	199039	100	Isovaleryl-CoA dehydrogenase deficiency	28940889
kissing spine, thoracic region	m4824	62	pollen-specific receptor-like kinase 3, arabidopsis	11942	57	Increased analgesia from kappa-opioid receptor agonist, female-specific	1805007 1805008
klippel-feil syndrome	q761	73	li-fraumeni syndrome 2	219002	100	Li-Fraumeni syndrome 2	63751220 78378222 35993958 35993958 55819519 55819519 28934574 28934574 28934574 28934576 28934576 11540652 11540652 11540652 11540652 28934573 17880604 17880604 35163653 1800372 1800372 28934578 28934578 28934875 28934875 28934873 28934873 55863639 55863639 1042522 1042522 1042522 1800371 1800371 1800370 17879961 17879961
knuckle pads	m721	61	ribavirin-5'-phosphate	21469	58	ribavirin response - Toxicity/ADR	1127354 7270101
kyphoscoliotic heart disease	i271	71	rh-null, regulator type	219698	72	Rh-null hemolytic anemia, regulator type	16879498
lamellar ichthyosis	q802	100	kid syndrome, autosomal recessive	198345	79	Autosomal recessive Dejerine-Sottas syndrome	28936682
late congenital neurosyphilis, unspecified	a5040	67	neutrophil-specific antigen na2	75662	93	Neutrophil-specific antigens na1/na2	448740
late-onset cerebellar ataxia	g112	76	migraine, familial hemiplegic, 1	197942	97	Familial hemiplegic migraine	28933401 28933400 28933398
lattice corneal dystrophy	h1854	68	cone dystrophy 4	221672	100	Cone dystrophy 4	71454844 200098356
legionnaires' disease	a481	76	complement component 5 deficiency	198032	88	Complement component 8 deficiency type 2	41286844 146187042
leigh's disease	g3182	93	cytochrome-c oxidase deficiency	198031	100	Cytochrome-c oxidase deficiency	119466000 1805124 140799936 28461189 28679680
lipoprotein deficiency	e786	65	intermediate density lipoprotein cholesterol	37739	62	High density lipoprotein cholesterol level quantitative trait locus 9	11591147 67608943 28362286 5935 41267007 5370 72552778 326 328 13702 120074117 1050239 28931573 1169288 183130 5882 7412 1127354 7270101 3843763
liver cell carcinoma	c220	80	carcinoma, renal cell	197910	88	Merkel cell carcinoma	2124437 11762213 11762213 34589476 34589476 11214077
liver cell carcinoma	c220	80	carcinoma, renal cell	197910	88	Merkel cell carcinoma	
long qt syndrome	i4581	100	long qt syndrome	198002	100	Long QT syndrome	119466000 72546668 72546668 72546668 72546668 28936686 45589741 45589741 41261344 41261344 41261344 7626962 7626962 7626962 45553235 45553235 45609733 72549410 72549410 45546039 72544141 72544141 36210421 36210421 41314354 28928904 28928904 28928904 28928904 731506 1800170 28730664 179489 179489 179489 179489 179489 17215479 17215479 28730754 1800171 1800171 1800171 1800171 1805118 12720449 12720449 12720449 17221854 17221854 17221854 17221854 34516117 1805128 1805128 1805128 1805128 1805128 17857111 28933384
lymphangitis	i891	61	glucosteroid response modifying factor	31020	68	corticosteroids response - Efficacy	1801131 1876828
lymphangitis	i891	61	glucosteroid response modifying factor	31020	68	corticosteroids response - Efficacy	
macular corneal dystrophy	h1855	70	cone-rod dystrophy 12	221279	98	Cone-rod dystrophy 2	61751408 1800728 61750126 61751374 61752410 61751392 61748550 61750200 137853907 10151259 28937883 28937877 72547544 61748436
magnesium deficiency	e612	100	familial hypomagnesemia - hypercalciuria	196916	85	Familial hypokalemia-hypomagnesemia	72550870 1801725 72552258 28936387
malignant ascites	r180	69	malignant hyperthermia	193511	100	Malignant hyperthermia	34934920 34934920 35364374 200563280 34390345 121918593
malignant hyperthermia due to anesthesia, sequela	t883xxs	69	malignant hyperthermia, susceptibility to, 1	196901	75	Hypertriglyceridemia, susceptibility to	4950928 4961 20541 2293869 2266788 2075291 3135506 2230500 1800566 5491 34934920 35364374 200563280 34390345 121918593 11909217
malignant mast cell tumor	c962	67	malignant variant of abrikosov's tumor	196783	69	Malignant tumor of urinary bladder	4962081

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
malignant melanoma of lip	c430	72	melanoma, familial	218290	70	Fatal familial insomnia	3731249 6413464 1800586 1126809 28897705 766173 11571640 28897706 41293475 56403624 28897708 28897709 28897710 11571653 28897715 2227943 28897717 28897718 55638633 28897720 28897722 28897723 55969723 28897726 28897727 28897728 41293491 28897731 56087561 11571657 41293497 41293497 55875643 4987117 28897737 1799954 56191579 56191579 34309943 35029074 35029074 55953736 55977008 28897744 28897745 28897747 41293511 41293513 28897749 11571747 28897753 28897754 28897754 11571769 28897755 28897756 45580035 41293521 28897758 28897758 28897759 61757642 11571831 11571833 861539 1805006 11547464 1110400 41293465 8176316 28897698 45553935 28897696 28897696 55770810 1799967 1799966 1799966 28897693 56158747 28897692 28897691 55815649 1800744 41293455 41293455 28897690 28897689 55639854 28897686 28897686 55930959 62625307 28897685 16942 4986852 16941 1800704 56321129 41286300 28897684 1800709 28897683 56082113 28897682 4986846 4986845 4986844 4986850 28897680 1800064 56039126 56012641 28897677 28897676 62625300 56128296 1799950 8176153 28897675 1799965 34545365 28897672 1800062 1799990 28933385
malignant neoplasm of abdomen	c762	71	desmoid disease, hereditary	197128	64	Hereditary diffuse gastric cancer	121965063 121965064 28934608 1801155 34919187 2229994 2229995 150766139 5030874 33935154 35572355
malignant neoplasm of cervix uteri, unspecified	c539	74	primary malignant melanoma of the cervix	197542	70	Malignant melanoma of skin	16891982
malignant neoplasm of parathyroid gland	c750	77	parathyroid cancer, adult	198175	73	Thyroid cancer, anaplastic	28934576
mandibulofacial dysostosis	q754	100	mandibulofacial dysostosis	196886	94	Mandibuloacral dysostosis	28928902 57318642 57629361 60580541 56673169 60890628 58596362
manganese deficiency	e613	71	glutamate formiminotransferase deficiency	198396	100	GLUTAMATE FORMIMINOTRANSFERASE DEFICIENCY	16879498 28931584 61754634 28938472 28941768
maple-syrup-urine disease	e710	100	maple syrup urine disease	196917	100	Maple syrup urine disease	12021720 12021720 12021720 74103423
mcardle disease	e7404	75	rippling muscle disease 2	196894	100	Rippling muscle disease 2	28936685 28936686
medium chain acyl coa dehydrogenase deficiency	e71311	64	5 alpha fluorouracil toxicity	193495	67	fluorouracil response - Efficacy, Toxicity/ADR	121434280 121434277 121434277 121434281 121434274 121434282 77931234 77931234 67376798 3918290 3918290 55886062 1801265 72549310 1801019 1042522
medullary cystic kidney	q615	82	medullary cystic kidney disease 2	207216	100	Medullary cystic kidney disease 2	28934584
megalencephaly	q045	100	obesity	197016	100	Obesity	2282440 2491132 28932472 57865060 9282671 4684677 696217 34911341 1805192 1800571 1042714 7732671 1044498 7754561 4994 2229707 17848368 1421085 5030980 52804924 2229616
melas syndrome	e8841	76	neuroacanthocytosis, mcleod type	219196	88	McLeod neuroacanthocytosis syndrome	28937582 28940571 28933690
melkersson's syndrome	g512	76	sjogren-larsson syndrome	197751	98	Sjögren-Larsson syndrome	72547554 72547562 72547564 72547569 72547571 72547575
merrf syndrome	e8842	83	marfan syndrome	193512	100	Marfan syndrome	61752334 72658163 363811 363835 363807 140649 140598 2228241 140587
metabolic encephalopathy	g9341	74	epileptic encephalopathy, early infantile, 3	218417	99	Epileptic encephalopathy, early infantile, 36	28940289 200086262 146925326
metabolic syndrome	e8881	63	or-1 metabolite	132901	63	Glipizide poor metabolizer	3816873 1057910
metachromatic leukodystrophy	e7525	100	leukodystrophy, metachromatic	218377	80	Metachromatic leukodystrophy, juvenile type	28940893 28940893 28940893 80338820 80338820 743616 74315479 74315479 74315479 74315479 2071421 28940894 28940894 74315472 74315472 80338819 74315457 74315457 74315457
methylmalonic acidemia	e71120	90	malonyl-coa decarboxylase deficiency	196909	96	Deficiency of malonyl-CoA decarboxylase	45590836 121918248 121918248 1799971 9593 28941784 35648932 28937908 104894528
microcephaly	q02	100	hoyeraal hreidarsson syndrome	197235	100	Hoyeraal Hreidarsson syndrome	33927012 33927012 3125630 3125630 3125630 7528827 7528827 7528827 36004306 36004306 36004306 36004306 36004306 36004306 3762271 3762271 3762271 12138336 12138336 12138336 41310927 41310927 41310927 1412640 1412640 1412640 964201 964201 964201 199422168 199422168 199422168 16841081 16841081 16841081 7551108 7551108 7551108 6428388 6428388 6428388 12025066 12025066 12025066 200976140 41291187 35062203 34094720 10262966 28931608 930557 930557 930557 1057090 1057090 1057090 1057091 1057091 1057091 769420 769420 34767364 34767364 61754966 61754966 61754795 61754795 61753720 61753720 180177366 149616199 104886457 104886456 36045913 2234584 34677591 34677591 11214077 11214077 202058504 202058504 202058504 182018947 182018947 182018947 1800124 17227403 17233141 121918161 121918161 55770810 4988346 28903098 28935177 61753968 61750240 28934906 28936072 28936072 28936072
microcystoid degeneration of retina, bilateral	h35423	63	retinitis pigmentosa 41	221433	100	Retinitis pigmentosa 41	137853907
microgenia	m2606	80	micropenis	197737	100	Micropenis	9332964
microphthalmos	q112	100	nanophthalmos 2	218961	100	Nanophthalmos 2	45598538 28937894 121908189

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
mild intellectual disabilities	f70	83	intellectual disability	196556	100	Intellectual disability	29001685 1801274 143038880 7626962 199884004 199884004 9289231 9289231 201330912 72544141 72556370 28936670 4988498 1800073 142540482 1800076 1800076 75961395 75961395 77834169 77834169 78655421 78655421 78655421 79660178 35516286 35516286 78756941 77188391 191456345 56093012 1800085 121909011 77409459 78909279 73215912 1800089 213950 1801178 1800095 1800095 76713772 113993959 121909005 75527207 1800097 1800097 1800098 1800100 121908760 121908810 1800103 80224560 79633941 121908797 75096551 149279509 1800112 78194216 78194216 1800114 1800114 1800114 36210737 36210737 75039782 76649725 76649725 11971167 11971167 80034486 36210421 1805120 35916840 56214919 2230806 29001665 179489 179489 12720449 12577654 11024034 1800172 6265 17215437 137852761 1800054 1800054 3218707 3218674 56123940 2235006 641252 1800056 3092857 3092842 1800058 1800889 1801673 1800061 1800061 28904921 55801750 55982963 55982963 17174393 17174393 1800558 138326449 140621530 28933383 28933383 17782839 28756987 28756986 115352435 138358708 138358708 7221086 1800470 28934891 121964962 5742905 28935171 72554634 72554636 72554639 72554640 72554644 72554644 72554645 72554649 72554650 72554652 28935468 61748421
mitochondrial metabolism disorder, unspecified	e8840	64	superoxide dismutase 2, mouse	130925	83	SUPEROXIDE DISMUTASE 2 POLYMORPHISM	4880
moderate protein-calorie malnutrition	e440	63	colorectal mutant cancer protein, human	71421	69	Colorectal / endometrial cancer	63750664 2020912
mononeuritis multiplex	g587	73	desmosterolosis	220833	100	Desmosterolosis	28939092
mucolipidosis iv	e7511	83	sialidase deficiency	198348	77	Ganglioside sialidase deficiency	193302855 104886461
mucopolysaccharidosis, type ii	e761	87	mucopolysaccharidosis iv	197393	88	Mucopolysaccharidosis, MPS-II	72555368 72555391 72555365 72555367 72555369 72555362 72555366 72555364 72555392 121965019 34946266 34946266 34159654 34159654 1141608
mucopolysaccharidosis, type ii	e761	87	mucopolysaccharidosis iv	197393	88	Mucopolysaccharidosis, MPS-II	
multiple myeloma in remission	c9001	80	m protein, multiple myeloma	86238	75	Multiple myeloma, resistance to	1805388 1805388 1805389 1805389
multiple sclerosis	g35	100	multiple sclerosis	220222	100	Multiple sclerosis	1800693
mumps encephalitis	b262	76	autoimmune diseases	193506	77	Autoimmune thyroid disease 3	180223
muscle carnitine palmitoyltransferase deficiency	e71314	92	carnitine palmitoyltransferase ii deficiency, late-onset	218782	100	Carnitine palmitoyltransferase II deficiency, late-onset	28936674 28936674 2229291 1799821 28936673 28936673 28936673 74315293 74315293 74315293
muscular dystrophy	g710	89	muscular dystrophies	196892	89	Muscular dystrophy	28940869 28940869 41313880 41313880 60864230 60864230 58912633 58912633 58912633 60682848 60682848 60682848 57207746 57207746 59332535 59332535 58048078 58048078 58048078 56771886 56771886 60458016 60458016 60458016 60458016 60458016 60458016 61672878 61672878 61672878 57508089 57508089 61094188 61094188 58932704 58932704 58932704 58932704 57629361 57629361 57629361 57629361 60934003 60934003 57496341 73991549 72546667 72546667 72546667 28936383 28936383 28937597 28937597 28940576 61755781 61755769 4738824 34006675 34006675 34006675 28941782 200056620 142027093 142027093 142027093 28940274 28940273 1805142 28941469 28941468 28933098 28999113 28933693 28933693 28937905 28937905 28937900 28937900 28937900 28937901 28937901 28937903 28937903 1800280 1800280 1800278 1800278 228406 228406
myelofibrosis	d7581	76	primary myelofibrosis	197908	76	Myelofibrosis	121913615 121913615 77375493
myoadenylate deaminase deficiency	e792	80	amp deaminase/deficiency	199098	87	Muscle AMP deaminase deficiency	35859650 17602729
myocardial degeneration	i515	79	goldmann-favre syndrome	219699	100	Goldmann-favre syndrome	28937873 28937873
myoclonus	g253	100	epilepsies, myoclonic	197330	78	Myoclonic epilepsy, juvenile 1	3804505
myotonia congenita	g7112	81	tamoxifen/adverse effects	196770	58	tamoxifen response - Efficacy, Toxicity/ADR	28936670 55960271 28933402 3892097
nausea	r110	73	otraspray	21867	67	Rasopathy	56219475 12628 2510152 17586159
neonatal adrenoleukodystrophy	e71511	91	adrenoleukodystrophy	197747	82	Leukodystrophy	72466451 61752138 35333334 34002892 34159654 28940893 80338820 743616 74315479 2071421 28940894 74315472 80338819 74315457
neonatal candidiasis	p375	71	familial chronic mucocutaneous candidiasis	198879	100	Familial chronic mucocutaneous candidiasis	16910526
neonatal diabetes mellitus	p702	84	permanent neonatal diabetes mellitus	218759	100	Permanent neonatal diabetes mellitus	2476601 231775 3755319 1805192 237025 41295061 11594656 5219 5219
nephrogenic diabetes insipidus	n251	95	diabetes insipidus, neurogenic	220123	78	Neurohypophyseal diabetes insipidus	28931580 28934878
nephroptosis	n2883	73	nephronophthisis 1	198633	94	Nephronophthisis	28940891 34248917 121918244 2070635 200844390 353630 3794109 61747071
nervousness	r450	63	epidermal nevus	231519	100	Epidermal nevus	11554290 28931614
neurofibromatosis, type 1	q8501	96	neurofibromatosis type 5	198366	96	Neurofibromatosis, type 1	17887014 17884859
neurofibromatosis, type 2	q8502	65	somatostatin type 2a receptor	93927	61	simvastatin response - Toxicity/ADR	4149056 1719247 1346268
neuronal ceroid lipofuscinosis	e754	97	ceroid lipofuscinosis, neuronal, 1	219771	98	Ceroid lipofuscinosis, neuronal, 11	137852695 28940569 119455955 119455955 28940573 104894385 28940280 63749877
neuropathic heredofamilial amyloidosis	e851	81	transthyretin amyloidosis	221689	78	Amyloidogenic transthyretin amyloidosis	121918080 121918070 121918085 76992529 76992529
niemann-pick disease type c	e75242	100	niemann-pick disease, type c	197276	98	Niemann-Pick disease type C1	120074117 120074117 120074117 140130028 28942106 28942108 28940897 1788799 55680026 1805081
nocturia	r351	71	charcot-marie-tooth disease, recessive intermediate a	219331	100	Charcot-Marie-Tooth disease, recessive intermediate A	28937906

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
nonfamilial hypogammaglobulinemia	d801	74	x-linked agammaglobulinemia	198384	100	X-linked agammaglobulinemia	145092287 10127939 41310709
non-ketotic hyperglycinemia	e7251	87	hyperglycinemia, transient neonatal	218406	72	Cyanosis, transient neonatal	121964974 34878913
old myocardial infarction	i252	91	myocardial infarction	219333	100	Myocardial infarction	5174 5174 909253 1041981 12316150 6046 1048990 5918
orf virus disease	b0802	84	interferon gamma, receptor 1, deficiency	196759	97	Interferon gamma receptor deficiency	17181471
organic oligospermia	n4611	75	follicle-stimulating hormone deficiency, isolated	198090	100	Follicle-stimulating hormone deficiency, isolated	10835638 5030776
osteochondrodysplasia, unspecified	q789	80	oto-palato-digital syndrome type 1	197233	94	Oto-palato-digital syndrome, type I	28939677 28936703 104893915 104893915 1131454 10774671 28936368 1800472 1800471 1800470 1800469 28935470 28935470 28935469
osteogenesis imperfecta	q780	100	osteogenesis imperfecta, type 1a	197209	91	Osteogenesis imperfecta type 1, mild	72659361 72659361 72656402 72656402 72656402 72658151 72658151 72658151 72658151 72658161 72658161 72658161 72658161 72658176 72658176 72658176 72658200 72658200 72658200 72659319 72659319 72659319 72659319 72659319 72659319 72659319 72659324 72659338 72659338 72659338 72656352 72656352 72656343 72656343 34940368 72656324 72656324 72656321 72656321 72656314 72656314 72656306 72656306 72653173 72653172 72653172 66523073 66523073 66523073 72653169 72653169 72653166 72653166 72653154 72653154 72653152 72653152 72653137 72653137 72653131 72653131 72653131 72651657 72651657 72651653 72651653 67368147 67368147 67368147 72648356 72648356 67682641 67682641 67682641 72648333 72648333 72648320 72645357 72645357 72645357 72645353 72645353 72645353 72645333 72645333 72645331 72645331 72645328 72645323 72667037 72667037 72667023 67828806 67828806 67828806
osteopetrosis	q782	88	cataract	196543	100	Cataract	2476601 28937595 231775 28931604 3792267 1805192 4402960 10010131 6446482 71524377 28937890 10946398 7756992 1044498 237025 3735231 72549406 72549406 72549405 35155575 200185429 13266634 41295061 11594656 7903146 12255372 5219 200203460 1131454 10774671 12926089 28934573 35910969 41282065 142090709
other abnormalities of breathing	r0689	75	spranger schinzel myers syndrome	197005	70	Gerstmann-Straussler-Scheinker syndrome	11538758
other absence of family member	z6332	62	lethal factor toxin susceptibility 1 protein, mouse	151942	65	Diabetes mellitus, ketosis-prone, susceptibility to	2229291 1799821 35155575
other adrenocortical insufficiency	e2749	76	acth resistance	220460	100	ACTH resistance	41525747 4988235 41380347 182549 1805015 28940892
other adrenogenital disorders	e258	69	dystonia, dopa-responsive, with or without hyperphenylalaninemia, autosomal recessive	198494	100	Dystonia, dopa-responsive, with or without hyperphenylalaninemia, autosomal recessive	45471299 28934580 28934581 6356 41298442
other alzheimer's disease	g308	82	alzheimer disease type 1	197688	100	Alzheimer disease, type 1	58973334 63750048 63749851 63750197 63750197 28936379 63749884 63750666 63750110 63750110 63750592 63750900 17125721 63749964 63750399 63750734 63750973 63750643 63750066 63751039 63751039 63750671 63750671 63750847 63750363
other biotin-dependent carboxylase deficiency	d81818	62	biotin holocarboxylase synthetase	32336	86	Holocarboxylase synthetase deficiency	28934602
other blood donor, stem cells	z52091	62	lutheran blood-group system	90787	69	LUTHERAN BLOOD GROUP POLYMORPHISM Lu(a)/Lu(b)	28399653
other cardiomyopathies	i428	84	cardiomyopathies	196803	87	Cardiomyopathy	57077886 28928903 58922911 28928900 59270054 41313880 60864230 59026483 61195471 28933092 61295588 60682848 57207746 59332535 11264444 56816490 56771886 56851164 59301204 60458016 61672878 534807 58932704 57629361 57629361 60934003 56984562 56984562 61444459 60890628 45466197 63750197 3766871 34967813 6716782 201273719 28933405 62636495 62636491 62636492 73991549 28936685 63750743 41261344 45546039 45620037 41276525 34768413 28938173 201103536 147622517 202196166 201306690 71579353 1417635 36212066 35078470 2856655 36211723 11570082 34580776 2228387 35404804 61001398 58386780 193922674 1046116 201754030 201728041 45464193 45544633 45451303 45478699 3218716 3218713 45516091 45614536 45578741 57019720 61157095 57758262 59616921 57536312 59296273 28940896 28940896 58597584 59856285 60723330 2230234 76992529 6060980 199896820 28935490 3134560
other cardiovascular syphilis	a5209	67	cardiovascular heat shock protein, rat	216305	66	Cardiovascular phenotype	4074536 41313880 538089 505058 56984562 61444459 3729547 3766871 34967813 3813250 3829747 3731752 3731749 3731748 747122 9808377 2366751 3829746 3731746 744426 10164753 12464787 2288570 2303838 2288569 16866412 13390491 2627043 2742347 2291310 2291306 4894048 12476289 1552280 10497520 1058261 34337334 1058284 73991549 1974763 72546667 72546668 2340917 45489199 1805126 41261344 9858585 1805125 7430407 1805124 41312433 45471994 45620037 41276525 6791924 6599230 72544141 3733617 1016835 2076299 9493627 6964587 1063242 1063243 36210421 1805123 1137617 1805120 35136134 61755997 2228645 10997975 7916821 7079481 1417635 1417635 2234962 1800170 28730754 12720449 1057128 34516117 11601907 13451 35078470 36211723 1800565 34580776 17215437 2270676 2228387 1275085 1544514 215976 35404804 61001398 139620148 3759236 193922674 1046116 141062252 365990 150885220 45516091 1071646 45578741 7221086 173135 1893963 2230234 2278792 1042769 1791235 1800458 76992529 1051421 35761929 3810510 1805128 1805127 2301558 202059967 5992495 11541479 1800278 72466590 1801187 16990264 28935490

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
other cerebral infarction	i638	86	cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy	219159	90	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy	28933698 28937321
other cerebral palsy	g808	82	sutherland-haan x-linked mental retardation syndrome	198690	67	Encephalopathy, neonatal severeMental retardation, X-linked, syndromic 13Rett syndrome	61750240
other cerebrovascular syphilis	a5205	62	walker-warburg syndrome	196846	100	Walker-Warburg syndrome	28937900
other chronic pancreatitis	k861	87	hereditary pancreatitis	198253	100	Hereditary pancreatitis	148954387 35877720 1800076 213950 1800094 1800095 113993959 75527207 1800103 11971167 1800130 6666
other cirrhosis of liver	k7469	77	genetic diseases, inborn	218282	89	Genetic prion diseases	59914820 61752334 121918165 56208331 4738824 200737038 119455955 16024 111436401 11538758 11538758 1799990 16990018 28933385
other conduct disorders	f918	69	pseudoxanthoma elasticum	218921	100	Pseudoxanthoma elasticum	72547524 28939702 63749823 63750759 63751325 2238472 72664233 63749796 72653744 72653706 60791294 28939701 63750459 72664209 72650700 72650700 72657698 6504649
other congenital deformities of chest	q678	81	oculodentodigital dysplasia	218620	100	Oculodentodigital dysplasia	28936670 28931601 17653265
other congenital functional disorders of colon	q432	71	congenital disorder of glycosylation type 1g	196951	100	Congenital disorder of glycosylation type 1G	35383149 201337954 28940588 28939087 28934876 28928906 28942090
other congenital ichthyosis	q808	84	ichthyosis prematurity syndrome	197413	100	Ichthyosis prematurity syndrome	137853131
other congenital viral diseases	p358	71	radiation tolerance	220025	67	Exercise intolerance	28931610 147014855
other corneal degeneration	h1849	71	retinitis pigmentosa 15	219497	100	Retinitis pigmentosa 15	61750641 61751408 61753034 61751407 1800728 62642574 61750152 61752435 62642573 61750130 61750120 61750064 61751374 61750061 1801581 58331765 61749438 61751263 61748557 61751392 61748550 61748548 61750200 146733615 35365413 62642057 62638651 62638644 62638637 62638634 62638632
other creutzfeldt-jakob disease	a8109	86	creutzfeldt-jakob disease, sporadic	219895	85	Jakob-Creutzfeldt disease	1799990 28933385
other cystic kidney diseases	q618	88	central nervous system diseases	196751	68	Spongy degeneration of central nervous system	28940891 199469707 199469707 199469707 199469707 3792267 4402960 151279194 151279194 151279194 147416429 147416429 147416429 10946398 7756992 1044498 137853108 199821258 200185429 13266634 200844390 7903146 12255372 5219 62640570 121918204 121918204 61747071 63751297 12948217 28940279 28940574
other disorders of aromatic amino-acid metabolism	e708	67	methylmalonyl-coa epimerase deficiency	220064	100	Methylmalonyl-CoA epimerase deficiency	111033538
other disorders of autonomic nervous system	g908	79	dopamine beta hydroxylase deficiency	196820	100	Dopamine beta hydroxylase deficiency	1611115 1108580
other disorders of iron metabolism	e8319	69	coumarin, poor metabolism of	218580	79	Debrisoquine, poor metabolism of	141982812 1128501 28399504 4986893 4244285 56337013 33950507 33930165 8177374 1801272 3892097 5030865 5030655 1065852
other disorders of phosphorus metabolism	e8339	87	phosphorus metabolism disorders	220134	63	efavirenz response - Metabolism/PK	776746 12248560 12248560 2279343 2279345 28399499
other disorders of purine and pyrimidine metabolism	e798	71	ribose-phosphate pyrophosphokinase/metabolism	221183	63	morphine response - Metabolism/PK	1799971
other disorders of tooth development	k008	77	leydig cell hypoplasia	218355	77	Islet cell hyperplasia	6688832 28936678
other disorders of urea cycle metabolism	e7229	66	drug metabolism, poor, cyp2c19-related	218964	58	Drugs used in opioid dependence response - Metabolism/PK	1799971
other epidermolysis bullosa	q818	88	kindler syndrome	197457	94	Kindler's syndrome	146180696 121918293
other fecal abnormalities	r195	71	sudden infant death	219073	81	SUDDEN INFANT DEATH SYNDROME	28928900 60864230 61214927 58672172 61282106 28928902 28928902 28928902 57920071 59981161 60890628 60890628 60890628 57830985 201091809 72551362 72551363 72551364 7626962 104893931 104893927 28931600 17653265 3734805 2046210 9397435 104894093 28933972 28933971 28933373 200389141 148969222 148969222 59616921 28940896 28936696 57521499 28935497 28358582 28358582
other frontotemporal dementia	g3109	88	frontotemporal dementia	218561	100	Frontotemporal dementia	63750818 63750818 63750653 63750653 63750355 63750355 63750652 63750652 63751048 63751048 63751006 63751006 63750331 63750331 63751243 63751243 63750077 63750077 63749801 63749801 63749877 63749877 63751294 63751294 63750349 63750912 63751273 63751165 63750972 63750092 63750570 63750424
other glycogen storage disease	e7409	85	glycogen storage disease iiia	221048	81	Glycogen storage disease II, adult form	113994126 113994126 113994130 113994131 192044702 121918419 202143236 202143236 202143236 1801175 1801176 80356484 28937909
other gm2 gangliosidosis	e7509	68	ganglioside gm3 lactone	53044	77	Gm2-gangliosidosis, late onset	72555371 72555373 72555391 72555391 72555391 72555359 72555361 72555366 72555366 72555366 72555360 72555392 72555392 72555392 72555392 72555358 9289231 28936670 2230806 138326449 140621530 28942072 121907954 28941771
other hemochromatosis	e83118	84	hemochromatosis, type 3	198241	100	Hemochromatosis type 3	28939076 1799945 1800730 28934597 28934596 1800562 1800562 41302357
other hemoglobinopathies	d582	71	hemoglobin inkster	128455	100	HEMOGLOBIN INKSTER	41331747
other hereditary corneal dystrophies	h1859	91	bietti crystalline corneoretinal dystrophy	196686	100	Bietti crystalline corneoretinal dystrophy	199476189
other hodgkin lymphoma, spleen	c8177	78	stomach neoplasms	196862	89	Neoplasm of stomach	28936699 28933375 35947132 3092857 3092857 35572355 28933369
other hydrocele	n432	65	butabarbital - hydrochlorothiazide - reserpine	188	73	hydrochlorothiazide response - Efficacy	2234922 61754278 7297610 16960228 4149601

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
other hyperparathyroidism	e212	75	thyrotropin deficiency, isolated	14686	88	Isolated lutropin deficiency	28942098 28942098 5030773
other hyperphenylalaninemias	e701	74	hyperphenylalaninemia, non-pku mild	221493	92	Hyperphenylalaninemia, non-pku	5030860 5030856 62642937 62516060 62514958 62642934 62517167 62507347
other hypertrophic cardiomyopathy	i422	90	cardiomyopathy, hypertrophic	197097	79	Cardiomyopathy, hypertrophic, mitochondrial	56851164 56851164 45466197 45466197 28933405 28933405 41261344 41261344 28938173 28938173 28938173 201103536 201103536 147622517 147622517 202196166 202196166 201306690 201306690 71579353 71579353 71579353 36212066 36212066 36212066 35078470 35078470 2856655 2856655 2856655 36211723 36211723 36211723 11570082 11570082 34580776 34580776 201728041 201728041 201728041 45544633 45451303 45451303 45478699 45478699 3218716 3218716 3218716 3218713 3218713 3218713 45614536 45614536 45578741 45578741 28937598 28937868 28935490 28935490 3134560
other inherited spinal muscular atrophy	g121	73	spinal muscular atrophy, distal, autosomal recessive, 4	221152	100	Distal spinal muscular atrophy, autosomal recessive 4	63750315
other intervertebral disc degeneration, lumbar region	m5136	76	intervertebral disc disease	196760	75	Intervertebral disc disease, susceptibility to	61734651
other intestinal obstruction	k5669	84	mngie disease	198441	86	Tangier disease	201330912 200440128 28937314 2853578
other keratitis	h168	75	keratitis-ichthyosis-deafness syndrome, autosomal dominant	197321	100	Keratitis-ichthyosis-deafness syndrome, autosomal dominant	72561723 28929485
other manic episodes	f308	60	peptide transport system	50153	69	PEPTIDE TRANSPORTER PSF2 POLYMORPHISM	241447 1800454
other mast cell activation disorder	d8949	66	leukocyte adhesion deficiency 3	221535	97	Leukocyte adhesion deficiency	143153871 2280965
other measles complications	b0589	73	diabetes complications	197184	70	Microvascular complications of diabetes 2	1799945 1800562 4880 1617640
other methemoglobinemias	d748	83	methemoglobinemia, congenital, autosomal recessive	198763	83	Congenital myotonia, autosomal recessive form	72552771 28931608 72552772 55960271
other mitochondrial metabolism disorders	e8849	68	mitochondrial complex i deficiency	198439	100	Mitochondrial complex I deficiency	142441643 11544803 11544803 63751061 776746 28939679 1801316
other muscle spasm	m62838	67	ankyrin repeat domain 2 (stretch responsive muscle) protein, mouse	152462	51	Alkylating Agents, anthracyclines and related substances, fluorouracil, and Platinum compounds response - Efficacy	1800566
other mycobacterial infections	a318	78	mycobacterial disease, susceptibility to, x-linked 1	221189	71	Autism, susceptibility to, X-linked 3	1676486 4833095 6046 2066844 2066845 5743289 2066847 61753973 61751444 61751370 61751362 61749705 61748414 28934906
other neurofibromatosis	q8509	80	pheochromocytoma	218829	100	Pheochromocytoma	33927012 34916635 5030824 34677591 11214077
other neutropenia	d708	79	fibrous dysplasia of bone	197087	82	Fibrous dysplasia of jaw	28936382 28937896 28938170
other night blindness	h5369	83	night blindness, congenital stationary, type 1a	197279	100	Congenital stationary night blindness, type 1A	201153410 62638625 62638624 62638214 62638208 62638202 62637021 62637037
other ocular albinism	e70318	84	albinism, ocular, type i	198781	100	Ocular albinism, type I	58933950 62635018
other oculocutaneous albinism	e70328	88	xanthism	198188	71	Autism	1858830 7794745 2710102 1861972 1861973 28940881 61753180 28940876 61754381 61754388 1126809 1126809 45474795 1805007 1805008 61753973 61751444 61751370 61751362 61749705 61748414 28934906
other peroxisomal disorders	e71548	88	peroxisome biogenesis disorders	197748	94	Peroxisome biogenesis disorder 7B	61752095 61752092 61750434 61752116 61752115 61753238 61750420 61752128 61752127 61752119 61752117 61752137 28936697 61752112 61752103 61752103 28936698 61752132 28940308 62641228
other porphyria	e8029	76	coproporphyria	196707	100	Coproporphyria	28931603
other primary ovarian failure	e2839	75	premature ovarian failure 3	219046	100	Premature ovarian failure 3	28937885
other proteinuria	r808	65	d-serine responsive transcript-1 protein, rat	128277	60	risperidone response - Toxicity/ADR	1137100 1137101 9282858 1045485 2952768 1670533 3796619 1045642 7493 2228063 1057910 1801203 1800497 28897728 55953736 11555096 28897696 28897676 28897675 28903098 489693 1047781 4680 3892097 28937312 2234036 28933688 1414334
other restrictive cardiomyopathy	i425	75	tight skin contracture syndrome, lethal	197966	100	Lethal tight skin contracture syndrome	58596362
other sideroblastic anemias	d643	85	anemia, sideroblastic, and spinocerebellar ataxia	197489	100	Anemia sideroblastic and spinocerebellar ataxia	72554634
other specified acute disseminated demyelination	g368	66	charcot-marie-tooth disease, type 1c	198882	100	Charcot-Marie-Tooth disease, type 1C	59101996 58982919 28928910 28928910 9282774
other specified myopathies	g7289	62	intranuclear rod myopathy	231532	78	Myopathy, centronuclear, 3	28928909 28928909
other thrombophilia	d6869	81	thrombophilia due to thrombomodulin defect	220509	100	Thrombophilia due to thrombomodulin defect	6025 5935 72550707 41348347 41348347
other urticaria	l508	75	familial dermographism	197705	75	facial dysmorphism	201213306 121918161
ototoxic hearing loss, bilateral	h9103	83	thrombocytopenia	196771	100	Thrombocytopenia	28928908 28941470 5918 5917
pallor	r231	67	palmoplantar keratoderma, nonepidermolytic, focal	199334	100	Palmoplantar keratoderma, nonepidermolytic, focal	59856285 60723330
paranoid schizophrenia	f200	63	schisphenin a	232700	71	Schizophrenia 4	2904552
paraphimosis	n472	79	connective tissue disorder, marden-walker type	197101	61	Sleep-wake schedule disorder, delayed phase type	28937884 28937884 28936679

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
parkinson's disease	g20	94	parkinson disease	198177	100	Parkinson disease	421016 421016 72470544 72470544 72470544 72470545 72470545 72470545 72554080 72554080 72554080 696217 283413 283413 1801334 1801334 1801582 1801582 56092260 56092260 34424986 34424986 1801474 1801474 55774500 55774500 6265 2256408 2256408 75054132 75054132 10878245 10878245 35517158 35517158 57355477 57355477 35847451 35847451 75711334 75711334 7308720 7308720 72546319 72546319 10878307 10878307 34410987 34410987 7966550 7966550 35808389 35808389 34805604 34805604 7133914 7133914 72546327 72546327 11175964 11175964 34995376 34995376 35507033 35507033 33958906 33958906 1427263 1427263 33949390 33949390 11176013 11176013 35303786 35303786 11564148 11564148 35801418 35801418 10878371 10878371 34015634 34015634 34637584 34637584 35870237 35870237 33995883 33995883 10878405 10878405 34869625 34869625 34778348 34778348 33962975 33962975 3761863 3761863 63751392 63751392 63751392 63751273 63751391 63750425 906807 906807 71799110 71799110 71799110 2853826
patent ductus arteriosus	q250	100	corneal diseases/congenital	196557	81	Central core disease	13216733 2817394 35364374 200563280 28933396 63749869
pectus carinatum	q677	62	lodenafil carbonate	193757	64	sildenafil response - Efficacy	5443
pedophilia	f654	67	factor viii, human	80291	78	FACTOR VIII (OKAYAMA)	4673 28935499
perianal venous thrombosis	k645	65	cardiac tropomyosin protein, xenopus	165149	68	Venous thrombosis, protection against	61754487 6048
periodic paralysis	g723	76	paralyses, familial periodic	221000	76	Periodic paralysis	28930069 28930069 2281845 2281845 17215437 62070884 62070884 200563280 200563280
periodontosis	k054	85	periodontitis, aggressive, 2	221092	96	Periodontitis, aggressive, 1	28937571
photokeratitis, bilateral	h16133	67	protein kinase g, mycobacterium tuberculosis	11441	89	Mycobacterium tuberculosis, protection against	1801274 3188996 8177374
pigmentary retinal dystrophy	h3552	72	beta-pdh	35772	74	G6PD BETICA	1050829 1050828
pilar cyst	l7211	63	palmitoyl-homocysteine	52180	68	Homocysteine, total plasma, elevated	1021737
pneumococcal meningitis	g001	78	invasive pneumococcal disease, recurrent isolated, 1	218928	71	Invasive pneumococcal disease, protection against	8177374
pneumonia due to escherichia coli	j155	69	utp-glucose-1-phosphate uridylyltransferase	118258	68	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase	111033661 111033681 111033686 111033728 111033737 2070075 2070075 111033773 2070074
polycystic kidney, adult type	q612	80	polycystic kidney disease, type 1	197460	91	Polycystic kidney disease, adult type	45503297 7766366 41273726 61889560
polycythemia vera	d45	100	polycythemia vera	219289	100	Polycythemia vera	77375493
polydipsia	r631	60	pcd-p(m)	36990	74	PI M(PROCIDA)	28931569
polyorchism	q5521	63	pf2-6 polymer	202479	61	FU1/FU2 POLYMORPHISM	13551
polytrichia	l683	62	polysorb (silica)	91691	63	SLC22A4 POLYMORPHISM	1050152
pompe disease	e7402	77	nose diseases	196687	71	No MEN2 disease	1799895 1799939 1800861 1800863
porphyria cutanea tarda	e801	100	porphyria cutanea tarda	220976	72	Porphyria cutanea tarda, susceptibility to	28936677 28936677 28936676 28936676 1800562 1800562
portal hypertension	k766	77	hypertension, diastolic, resistance to	219096	75	Parkinson disease, resistance to	41511344 2853826
posterior cerebral artery syndrome	g462	61	apparent mineralocorticoid excess	198393	100	Apparent mineralocorticoid excess	28934592 28934594
posterior cord syndrome	g8383	68	progeria syndrome, childhood-onset	221605	78	Hutchinson-Gilford progeria syndrome, childhood-onset	60864230 60652225
postmenopausal bleeding	n950	60	modifier of snc1, 11, arabidopsis	214204	59	Body mass index, modifier of	1805192
posttransfusion purpura	d6951	100	posttransfusion purpura	218506	100	Posttransfusion purpura	5918 5917
potter's syndrome	q606	74	pettigrew syndrome	196978	100	Pettigrew syndrome	80338851 59912467 104894739
precordial pain	r072	67	abrin precursor	72759	73	Recurrent abortion	6025
prediabetes	r7303	63	p(bs-co-ba) polymer	146170	51	KIDD BLOOD POLYMORPHISM Jk(a)/Jk(b)	1058396
presbyopia	h524	63	myopia, susceptibility to	197265	79	Favism, susceptibility to	1801131 324420 2476601 61816761 421016 1801274 1801274 5368 763110 1051740 1051740 699 28932472 231775 17235409 4124874 696217 34911341 6280 279871 279845 279836 1801394 20541 2303067 2303067 2303067 1042713 1042713 1042714 1800562 2736191 1051931 1051931 1805018 1805018 662 2491397 2184026 3750344 3793784 35947132 2234767 6265 569108 569108 497116 1800693 16910526 11544238 671 1805097 1048661 3825942 1801275 1421085 1024611 5918 6267 1050828
primary adrenocortical insufficiency	e271	67	pigmented nodular adrenocortical disease, primary, 2	220774	100	Pigmented nodular adrenocortical disease, primary, 2	76308115
primary carnitine deficiency	e7141	100	hyperammonemia	197849	97	Hyperammonaemia	2631367 2631366 13180043 72552724 72552725 72552726 72552728 274558 274557 72552729 72552729 72552730 72552732 1045020 72554356
primary hyperparathyroidism	e210	100	parathyroid adenoma, familial	219270	69	Parathyroid adenoma, somatic	28931612
primary hypersomnia	f5111	80	hyperoxaluria, primary, type ii	197540	98	Primary hyperoxaluria, type III	34116584 34116584 34116584 34664134 11817730 11817730 11817730
primary open-angle glaucoma, left eye, mild stage	h401121	73	glaucoma, primary open angle	218450	100	Primary open angle glaucoma	9282671
primary pulmonary hypertension	i270	85	pulmonary arterial hypertension, hereditary hemorrhagic telangiectasia-related	218714	98	Pulmonary arterial hypertension related to hereditary hemorrhagic telangiectasia	28936687 28936688 28936401
progressive diaphyseal dysplasia	q783	63	epilepsy, progressive myoclonic 3	221205	97	Epilepsy, progressive myoclonic 6	28940576 63749877 141554661

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
progressive external ophthalmoplegia, bilateral	h4943	84	ophthalmoplegia, chronic progressive external	218997	90	Progressive external ophthalmoplegia	28999114 28999114 28937887 28937887 41549716 41549716
progressive vascular leukoencephalopathy	i673	69	leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation	221138	100	Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation	142433332 142433332
prolonged pregnancy	o481	76	prl-like protein d	101079	63	G6PD SAPPORO-LIKE	72554664
prolymphocytic leukemia of t-cell type, in remission	c9161	71	t cell acute-lymphocytic leukemia protein, rat	232073	72	T-cell prolymphocytic leukemia	28904921
propionic acidemia	e71121	72	glutathione synthetase deficiency	197898	99	Gluthathione synthetase deficiency	1799971 61749895 28936396
prostatosis syndrome	n4282	79	diarrhea	197707	89	Diarrhea 7	104893934 76163360 148665132
protein deficiency anemia	d530	65	breast cancer type 2 susceptibility protein, human	210451	66	Cancer of multiple types, susceptibility to	28997576 1799899 1801155 1800054 28904921 1800470 72552322 17879961 17879961
pruritus ani	l290	61	down-regulated in adenoma protein, mouse	155599	57	Colonic adenoma recurrence, reduced risk of	2302615
pruritus scroti	l291	61	prepro-neuropeptide y	48541	75	NEUROPEPTIDE Y POLYMORPHISM	16139
pseudohypoparathyroidism	e201	74	thyrotropin-releasing hormone resistance, generalized	220502	82	Thyroid hormone resistance, generalized	28933408
ptosis of breast	n6481	69	breast cancer, familial	218495	94	Familial cancer of breast	1045485 28997576 28997576 1801155 1800054 28904921 3092989 9567552 1799943 1799943 1799943 1799943 206120 4987046 2126042 2126042 2126042 28897705 766173 766173 11571640 28897706 28897706 28897706 144848 144848 41293475 41293475 56403624 56403624 28897708 28897708 28897709 28897710 28897710 28897710 11571653 28897715 2227943 28897717 28897718 28897718 1799944 55638633 55638633 41293477 41293477 28897719 28897719 1801406 1801406 1801406 28897720 28897722 28897723 28897725 28897725 55969723 55969723 28897726 28897727 28897728 41293491 28897731 56087561 56087561 28897735 11571657 11571657 41293497 41293497 55875643 55875643 55875643 4987117 28897737 1799954 56191579 56191579 34309943 35029074 35029074 35029074 55953736 55953736 28897742 28897742 206079 11571684 206081 11571686 9943876 55977008 55977008 4986859 28897744 9534262 9534262 28897745 28897746 28897746 28897747 41293511 41293513 41293513 41293513 41293513 28897749 28897749 28897749 28897749 28897750 9634672 28897751 28897751 11571747 28897753 4942486 28897754 28897754 11571769 28897755 28897755 28897755 28897756 45580035 45580035 11571789 206147 11571805 41293521 28897758 80359205 80359205 28897759 61757642 517118 11571831 11571831 11571831 11571833 11571833 28897761 28897761 28897761 15869 7601 12911192 6496742 2301826 35623 35625 4148350 4148351 35628 4148353 4148356 11075295 249954 8053188 1800566 28363284 8176320 8176319 8176318 41293465 41293465 41293465 8176316 8176316 1800751 1800751 28897698 28897698 3092988 8176289 45553935 45553935 45553935 8176273 8176265 41293461 41293461 41293461 41293461 3092994 3092994 45444999 45444999 28897696 28897696 55770810 55770810 55770810 8176258 1799967 1800726 1800726 1799966 1799966 1799966 1799966 28897693 56158747 56158747 28897691 28897691 28897691 55815649 55815649 1800744 1800744 8176199 3737559 4986849 4986849 41293455 41293455 41293455 1060915 1060915 28897690 28897690 799916 799916 28897689 55639854 28897686 28897686 28897686 55930959 55725337 55725337 62625307 16942 16942 55909400 55909400 4986852 16941 16941 1800704 1800704 56321129 799917 799917 799917 799917 799917 41286300 41286300 28897684 1800709 28897683 28897683 56082113 56082113 28897682 62625306 62625306 62625306 16940 16940 41286296 41286296 4986846 4986846 4986845 4986844 4986850 4986850 4986850 28897680 1800064 1800064 56039126 56039126 56012641 56012641 28897677 28897676 28897676 28897676 62625300 62625300 56128296 56128296 1799950 8176153 28897675 1799965 799923 799912 28897672 28897672 28897672 28897672 28897672 8176109 8176104 8176103 1800062 1800062 1800062 1800062 8176092 8176091 3765640 3092986 11655505 4793204 4988346 28903098 28903098 1800470 72552322 72552322 17879961 17879961 28909982
pulmonary mycobacterial infection	a310	81	disease susceptibility	197255	83	Prion disease, susceptibility to	2476601 6025 7775 288326 1801278 3948464 3948464 2114592 2114592 3856806 1049296 1800562 1044498 1169288 2073711 28914832 1024611 1024611 28933368 1799990
pulp degeneration	k042	81	macular degeneration, age-related, 11	221455	100	Age-related macular degeneration 11	1800728 61749423 800292 1061170 1410996 3732378 3732379 9332739 9332739 547154 547154 4151667 4151667 641153 641153 11200638 2230199 1064039 1064039
pulp degeneration	k042	81	macular degeneration, age-related, 11	221455	100	Age-related macular degeneration 11	
pyridoxine deficiency	e531	62	thyroxine-binding globulin deficiency	219201	90	Thyroxine-binding globulin deficiency, partial	121912708 1799971 1815739 28933689
rectal polyp	k621	62	tli polymerase	73602	65	TLR4 POLYMORPHISM	4986790 4986791
re-entry ventricular arrhythmia	i470	61	periventricular laminar heterotopia	207597	87	X-linked periventricular heterotopia	201273719 63750743 63750743 193922674 193922674 1046116 28935470
refsum's disease	g601	93	refsum disease	196749	81	Norum disease	28939672 28940887
renal osteodystrophy	n250	77	cone-rod dystrophy 6	199219	95	Cone-rod dystrophy	61751408 1800728 61750126 61751374 61752410 61751392 61748550 61750200 137853907 71454844 200098356 200311463 200311463 10151259 28937883 28933695 28933695 61750173 61750173 61750174 61750174 61748436
resistance to antiviral drug(s)	z1633	67	warfarin sensitivity	221195	68	warfarin response - Toxicity/ADR	7900194 1057910 9923231

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
restless legs syndrome	g2581	62	rubinstein-taybi syndrome	197071	100	Rubinstein-Taybi syndrome	6746030 41268673 28937315
retained plastic fragments	z182	70	soymorphin-5	224303	63	C3S/C3F POLYMORPHISM	201213306 1800255 121918161 2230199
retinal edema	h3581	74	basal laminar drusen	218578	100	Basal laminar drusen	1061170
retinal telangiectasis, bilateral	h35073	72	ataxia-telangiectasia variant	221030	100	Ataxia-telangiectasia variant	137852761 28904921
rett's syndrome	f842	93	rett syndrome	219209	100	Rett syndrome	72658163 62653623 62653623 62653623 62641235 62641235 62641235 62641235 62641235 61749700 61749700 61749700 61749704 61753251 61753251 61753251 62621672 61753979 61753978 3027927 63749064 3027928 61753965 61753016 61753000 61753000 61752361 61752361 61751449 61751448 61751444 61751443 28935468 61751441 61751440 61751439 61749723 61749723 61749723 61751370 61751367 61751362 61751360 61750259 61750249 63749012 61750247 61750245 61750242 61750240 1042870 61749721 61750232 61750231 61749750 61749749 61749743 61749739 61749736 61749715 61749715 61749734 61749729 61749727 61749726 61749720 61748381 61749718 61749717 61749714 61749709 61749708 61749707 61749703 61748427 61748425 61748421 61748421 61748420 61748418 61748417 61748417 61748416 61748415 28934906 61748408 61748407 61748407 28934905 28934905 61748406 61748404 61748402 61748400 61748399 61748398 61748396 61748396 61748395 28934908 61748393 61748390 61748390 28934904 28934904 28934904 61748384 61753982 61755763 61755763 61755761 61754459 61754457 61754457 28934907 28934907 61754455 61754453 61754453 61754453 61754452 61754451 28935168 61754438 61754428 61754426 62641234 61754425 63749008
reye's syndrome	g937	93	leiomyomatosis	196748	76	Lymphangiomyomatosis	4962081 35660529 45517126 45517130 45517139 45517148 45457694 34012042 45517185 45517189 45480292 13337626 45477491 45517299 45517309 35986575 45483392 45517423
rheumatoid arthritis, unspecified	m069	77	arthritis, rheumatoid	197752	100	Rheumatoid arthritis	2476601 3766379 2004640
rhinophyma	l711	60	chinophene	218	70	Schizophrenia	1801131 6280 2904552 6267
rhizomelic chondrodysplasia punctata	e71540	100	rhizomelic chondrodysplasia punctata, type 1	193490	100	Rhizomelic chondrodysplasia punctata type 1	11558492 11558492 61753238 61753238 1805137 1805137
ross river disease	b331	75	phosphopyruvate carboxylase deficiency	197741	90	Pyruvate carboxylase deficiency	28940589
sandhoff disease	e7501	100	sandhoff disease, juvenile type	219694	100	Sandhoff disease, juvenile type	820878 820878 820878 28942073 28942073 28942073
sarcoid arthropathy	d8686	63	pth-biotin	53821	64	British HPFH	35710727
scleritis with corneal involvement, bilateral	h15043	62	muscular dystrophy, congenital progressive, with mental retardation	199060	73	Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B5	28937900 28937901 28937903
sclerodactyly	l943	62	brachydactyly type c	198110	100	Brachydactyly type C	28933082 28933082 28928891 28928891 28936683 28936397
scorbutic anemia	d532	69	glutathione synthetase deficiency of erythrocytes, hemolytic anemia due to	220171	100	Glutathione synthetase deficiency of erythrocytes, hemolytic anemia due to	149616199 104886457 104886456 1800124 17233141 55770810 28931584 4988346 28903098 28938472
scrotal transposition	q5523	62	tonicity-responsive enhancer binding protein, human	115654	60	antipsychotics response - Toxicity/ADR	3753380 67376798 3918290 3918290 55886062 55886062 6025 2228001 1801019 1801019 6295 17244841 730012 746647 1800629 1800629 4713916 4713916 4713916 20455 1045642 1045642 121909005 75527207 6977820 12248560 1057910 1057910 1057910 1695 1800497 1800497 4149015 4149056 28940310 28940309 7294 9934438 9923231 17782313 489693 489693 489693 489693 2108622 25487 3212986 3212986 4680 3892097 3892097 3892097 3892097 3813929 1414334
secondary mast cell activation	d8943	67	sendai activating enzyme	44399	72	Enzyme activity finding	662 705379
sensorineural hearing loss, bilateral	h903	84	deafness, nonsyndromic sensorineural, mitochondrial	198766	100	Deafness, nonsyndromic sensorineural, mitochondrial	145191476 80356595 80356595 80356590 80356590 118203989 118203989 28942096 28942096 28942097 28942097 63749888 25640 28936670 61750427 116900495 116900495 116900495 60284988 60284988 111033199 111033199 36039758 80338849 80338849 111033244 111033244 111033312 111033312 111033312 111033312 121908362 121908362 151001642 151001642 55947360 55947360 146405172 72558199 72558200 34937870 1126809 121909305 121909305 3751385 3751385 28931595 28931593 28931593 28931593 28931593 72561723 72561723 72474224 72474224 2274084 2274084 121908866 45474795 201257588 28936682 5918 5917 28999112 28999111 28940307 16996652 11090865 3888511
septo-optic dysplasia of brain	q044	82	growth hormone deficiency with pituitary anomalies	221582	100	Growth hormone deficiency with pituitary anomalies	28936704
shared psychotic disorder	f24	74	aniridia	7024	100	Aniridia	2234584
short chain acyl coa dehydrogenase deficiency	e71312	72	2-methyl-3-hydroxybutyric aciduria	197244	100	2-methyl-3-hydroxybutyric aciduria	147936696 61732144 1800556 28940874 28941773 28940872 2230178 2230180 62626305 28935476
sick sinus syndrome	i495	78	sick sinus syndrome 1, autosomal recessive	219104	100	Sick sinus syndrome 1, autosomal recessive	45620037 45620037
siderosis	j634	78	diffuse mesangial sclerosis	198329	100	Diffuse mesangial sclerosis	28937590 28941778
siderosis	j634	78	diffuse mesangial sclerosis	198329	100	Diffuse mesangial sclerosis	
smith-lemli-opitz syndrome	e7872	76	nevus, pigmented	197546	63	White sponge nevus 2	80338864 80338862 61757582 80338859 138659167 80338857 80338856 80338855 28938174 60440396 59897026
snoring	r0683	71	efavirenz, (r)-isomer	98078	72	Efavirenz response	3745274 3745274 36079186 2279343 2279345 28399499 34097093

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
somatization disorder	f450	74	skin-hair-eye pigmentation, variation in, 9	221203	100	Skin/hair/eye pigmentation, variation in, 9	16891982 26722 17244841 12203592 35264875 35264875 3829241 3829241 1042602 1126809 1126809 12821256 12896399 1800407 1800401 12913832 1667394 1426654 148438059 1805005 2228479 2228479 1805007 1805008 6058017
sparganosis	b701	60	drevogenin i	66863	64	PI NULL(DEVON)	11558261
spinal muscular atrophy, unspecified	g129	79	neuronopathy, distal hereditary motor, type v	218771	94	Distal hereditary motor neuronopathy type 2B	63750315 57965306 28936686 121909192 121909192 28937597 28937568 28937568 28937569 28937569
spirillosis	a250	83	aspergillosis	196651	69	Legionellosis	2072493 5744168
spondyloepiphyseal dysplasia	q777	84	spondyloepiphyseal dysplasia with congenital joint dislocations	196993	100	Spondyloepiphyseal dysplasia with congenital joint dislocations	28937593
squamous cell carcinoma of anal skin	c44520	78	esophageal squamous cell carcinoma	218437	89	Esophageal squamous cell carcinoma, somatic	41341748 28937897
stannosis	j635	63	peg-gly-paclitaxel	111928	65	paclitaxel response - Efficacy	1056836 1042522
steroid responder, bilateral	h40043	63	serum response factor	127323	65	salmeterol response - Efficacy	1042713 1042713 7793837 776746 121909005 75527207 6988229 1799978 1876828
subacute sclerosing panencephalitis	a811	100	polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	197462	100	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	28939079 201258663
tay-sachs disease	e7502	100	tay-sachs disease	219664	100	Tay-Sachs disease	147324677 28940871 76173977 121907954
tetralogy of fallot	q213	100	tetralogy of fallot	197611	100	Tetralogy of Fallot	28936670 56208331 28374544
thanatophoric short stature	q771	68	thanatophoric dysplasia, type 2	197616	97	Thanatophoric dysplasia type 1	28933068
thoracic aortic aneurysm, ruptured	i711	84	familial thoracic aortic aneurysm and dissection	218492	83	Thoracic aortic aneurysm and aortic dissection	7579903 1800255 34833812 35766612 35766612 2228048 2291628 190450 32209 154001 1815739 363811 363835 140649 140630 140598 2228241 140587 55831697 2075511 16967494 2272554 4781689 2235491 1801181 2228298 5742905 234706
thrombocytopenia, unspecified	d696	73	thrombocytopenia 3	221487	86	Thrombocythemia 3	28928908 35948326 77375493 77375493
thyrotoxicosis factitia with thyrotoxic crisis or storm	e0541	72	thyroid carcinoma with thyrotoxicosis	220711	90	THYROID CARCINOMA WITH THYROTOXICOSIS, SOMATIC	28937584
tinea pedis	b353	67	tear proteins	48863	62	Tegafur response	67376798 3918290 55886062 1801019
tinea unguium	b351	67	titanium(iv) transferrin	114740	58	Transferrin serum level quantitative trait locus 2	1800562
toxic encephalopathy	g92	86	hepatic encephalopathy	219031	78	Encephalopathy	2229291 1799821 142433332 201330912 28939079 201258663 201754030 28933698 28937321 28940289 200086262 200086262 16990018 11568188 62653623 62653623 62641235 62641235 61749700 61749700 61749704 61749704 61753251 61753251 146925326 146925326 61753971 61753968 61750240 28934906
transcobalamin ii deficiency	d512	80	corticosteroid-binding globulin deficiency	219902	100	Corticosteroid-binding globulin deficiency	28929488
transient tic disorder	f950	62	pentacaine, trans-(+,-)-isomer	22577	64	pentazocine response - Dosage	2952768
trichomonal prostatitis	a5902	60	peginterferon alfa-2a	99950	66	peginterferon alfa-2b response - Toxicity/ADR	3832043 4148323 7270101
trimethylaminuria	e7252	100	trimethylaminuria	197659	87	Trimethylaminuria, mild	2266782 2266782 1736557 61753344 2266780 2266780
trisomy 21, translocation	q902	61	2-hydroxy-atorvastatin	181474	64	atorvastatin response - Toxicity/ADR	4693075
tropical sprue	k901	65	biofluoride 12	158147	76	BCHE, fluoride 2	28933390

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
tuberous sclerosis	q851	100	tuberous sclerosis	197461	80	Tuberous sclerosis syndrome	4962081 11243931 10901220 45517093 45517091 45481400 45517092 45517094 45460096 45517096 45517096 45517097 45517097 35660529 35660529 45517098 45517098 45485999 45485999 45517099 1051616 45517100 45517100 45517102 45503094 45517103 45517104 45517106 45517107 45517108 45517109 45488500 45481496 45473598 45473598 45517111 45470502 45505405 45505405 45517112 45517113 45517114 45517115 45460895 45517116 45468592 45517117 45506197 45498496 45488893 45488893 45460197 45517119 45517121 45517122 45443205 45517123 45473698 45473698 45517124 45517126 45517126 45517126 45517130 45517130 45517131 45468594 45468594 45466296 45466296 45442896 45442896 45453000 45517135 45517133 45489591 45517134 45506396 45506396 1800748 1800748 45517136 45517137 45517138 45517139 45487497 45517141 45517141 1131825 45458600 45458600 12927333 45517150 45517152 45484892 45484892 45517144 45517144 45517145 45517145 45468201 45451295 45469896 45517147 45517148 45517148 1800725 1800725 1800725 45517149 1800742 1800742 45469099 45458694 45475793 45517154 45517154 45517154 45517155 45517155 45457701 45517157 45517158 45517159 45517159 45509697 45509697 45517161 45517160 45478593 45478593 45493394 45517162 45517162 45484298 45484298 45515894 45515894 45443091 45491095 45517164 45517168 45446697 45517165 45517169 45517169 45517170 45517170 45494392 45481199 45481199 45514794 45517172 45517172 45460191 45517173 45447093 45517174 45517174 45517175 45517176 45517177 45517178 45517178 45457694 45457694 34012042 45517180 45517180 45517182 45517182 45517182 45517183 45517181 45477195 45517185 45517187 45469702 45517188 45517188 45471596 45517190 45464995 45517189 7187438 45517195 45517194 45517197 45517198 45517199 45517200 45517201 45509791 45509094 45509094 45517202 45517203 45517203 45517203 45480292 45454398 45481105 45492397 45492397 45437797 45509392 45491895 45488199 45517208 45517208 45488191 45458592 45517209 45483301 45486196 45517210 45517211 45501596 45517212 45517212 45517213 45517213 45517215 45517215 45517216 45517217 45464093 7196184 45517219 45517218 45517220 45517221 45517221 45517222 45517222 45517223 45517223 45517224 45509500 45499191 45517225 45517228 45517230 45517229 45517229 45481704 45517231 45485599 45517232 45517232 45517233 45517234 45517235 45489791 45485395 45517236 45517237 45517238 45517238 45517239 45509000 45517240 45517245 45517241 45517242 45517242 45517243 45517244 145738496 13331451 45517247 45517248 13337626 45517250 45517251 1800715 45483797 45463498 45476100 45476100 45489200 45475594 45517253 45470695 45517254 45517256 45517257 45517259 45517259 45517260 45517261 45517266 45517265 45517264 45517267 45517267 45517268 45517269 45517270 45517271 45517272 45517272 45517272 45517273 45517275 45438592 45471697 45514100 45514100 45517279 45438192 45517280 45465195 45491698 45491698 45464800 45464800 36078782 36078782 36078782 36078782 45487103 45517283 45517282 45517284 45517284 45517285 45517286 45517286 45517287 45517288 45517289 45517289 45517291 45478393 45517292 45517293 45451497 45451497 45505895 45505895 45487691 45487691 45517294 45517294 45517294 45477491 45448801 45448801 45517297 45483391 45517298 45438205 45438205 45517299 45517301 45517301 45467194 45467194 45517300 45517303 45517304 45462194 45462194 45482795 45482795 45517306 45486193 45486193 45517308 45517308 45517309 45517310 45517311 45517312 45480591 45517314 45478595 45478595 45446700 45448994 45517316 45517316 45517315 1800705 1800724 45517318 45517317 45517320 45517320 45517322 45517323 45517324 45517325 45462593 45517326 45468491 45468491 45517330 45466399 45466399 45517333 45517333 45438898 45438898 45449094 45474795 45507198 45517334 45487992 45487992 45517335 45517335 45517335 45517336 45448791 45517340 45517340 45517341 45517341 45517342 45517343 45446594 45517344 45517344 45491597 45517345 45454201 45501793 45471791 45471791 35986575 35986575 45497598 45517350 45482793 45517351 45482792 45496499 45517352 45514391 45517353 45495796 45517354 45517354 45488595 45517355 45473296 45514095 45517356 45517357 45517360 45517361 45517362 45517363 45517364 45517364 45517365 45478894 45517366 45437192 45517367 45503995 45517368 45511204 45517369 45517370 45455296 45517375 45517373 45493793 45517376 45517378 45514993 45446901 45482398 45485092 45517379 45517380 45517380 45517381 45517381 45471896 45501091 45517382 45517383 45511393 45517385 35534817 45457095 45457095 45500595 45483392 45483392 35118875 35118875 45475501 45475501 45517388 45445199 45445593 45445593 45517390 45484794 45487291 45498401 45474691 45517391 45517391 45517392 45517392 45517394 45517396 45482604 45517401 45506600 45517407 45517408 45517409 45517410 45517411 45517413 45517414 45517417 13332222 45477298 45517419 45517419 45517420 45517421 45506695 45517423 45517423 45517423 45455398 45486402 45498900
tumor lysis syndrome	e883	82	torg syndrome	197219	77	Muir-Torré syndrome	63750047
type 1 diabetes mellitus with diabetic dermatitis	e10620	80	mitochondrial diseases	197392	100	Mitochondrial diseases	10010131 6446482 2229707 17848368 3087374 2307440
type ab blood, rh negative	z6731	60	abo blood-group system	101827	100	ABO blood group system	8176747 1053878

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ICDM name	ICDM ID	ICDM score	MESH name	MESH ID	MESH score	Phenotype	RS ID
tyrosinase negative oculocutaneous albinism	e70320	100	oculocutaneous albinism, tyrosinase-negative	198660	100	Tyrosinase-negative oculocutaneous albinism	28933371 1799989 28940881 28940881 61753180 61753180 28940879 61753185 28940876 28940876 28940877 61753190 61754360 61754361 1042602 61754363 61754365 61754367 61754368 61754375 61754380 61754381 61754381 61754387 61754388 61754388 62645917 1126809 61754399
tyrosinemia	e7021	96	4-alpha-hydroxyphenylpyruvate hydroxylase deficiency	197046	100	4-Alpha-hydroxyphenylpyruvate hydroxylase deficiency	17860424 1154510
underdosing of cannabis (derivatives), sequela	t407x6s	67	ipg cpd	129543	63	G6PD KAIPING	67376798 3918290 55886062 1801274 1801019 1049296 35378915 60554162 61145796 28933087 59685571 58556099 199422211 11558261 1042522 72554664
unspecified corneal degeneration	h1840	65	cerebellar degeneration-related antigen, human	67801	53	appendicular lean mass relative to body height	4646450
unspecified macular degeneration	h3530	85	macular degeneration, age-related, 2	218330	97	Age-related macular degeneration 8	61750130 1801581 61748550 3732378 3732378 3732379 3732379 3793784 10490924
unspecified malaria	b54	61	spectrin cagliari	85223	67	G6PD CAGLIARI	5030868
vanadium deficiency	e616	76	guanidinoacetate methyltransferase deficiency	198571	97	Deficiency of guanidinoacetate methyltransferase	17851582 55776826 80338734
ventricular tachycardia	i472	100	tachycardia, ventricular	197467	100	Ventricular tachycardia	61094188 61094188 2873479 2873479
viral endocarditis	b3321	61	2c-like protein, norwalk virus	83449	64	Norwalk virus infection, resistance to	601338
visual hallucinations	r441	65	arterial calcification, generalized, of infancy	198407	98	Generalized arterial calcification of infancy 2	28933977 63750759 63750759 72653706 72653706 72664209 72664209 72650700 72650700
vitreoretinal dystrophy	h3551	81	retinal dystrophies	197446	89	Retinal dystrophy	61751383 61751383 1800728 61751404 61750135 199476189 61755802 61755789 61755786 61755781 113624356 61752067
vomiting without nausea	r1111	63	hemiplegic migraine, familial type 2	198239	100	Familial hemiplegic migraine type 2	28933401 28933400 28933398
von willebrand's disease	d680	98	pseudo-von willebrand disease	197574	85	von Willebrand disease type 2N	61751310 61750630 61750612 61750612 61750584 61750581 61750117 61750101 61749403 61749398 61749398 61749397 61749392 61749387 61749384 61749380 61749380 61749372 61748497 61748497 41276738 41276738 41276738 41276738 61748478 61748478 61748477 61748477 61754011 61754010 61754002 61754002
weakness	r531	75	nonsyndromic deafness	231540	82	Non-syndromic genetic deafness	111033455 200147906 80356590 118203989 151001642 28931593 72474224 72474224 72474224 148690740
white piedra	b362	61	ziprasidone hydrochloride	92631	59	ziprasidone response - Toxicity/ADR	489693 489693
wilson's disease	e8301	80	spongiform encephalopathy with neuropsychiatric features	219591	100	Spongiform encephalopathy with neuropsychiatric features	36053993 35352891 34612342 3219484 60986317 2282057 60431989 1801249 76151636 1801248 1801246 72552255 732774 28942076 1061472 137853282 137853283 28942074 28942075 72552285 1801244 1801243 16990018
xeroderma pigmentosum	q821	100	xeroderma pigmentosum, group b	218367	97	Xeroderma pigmentosum, group F	56307355 1800124
zellweger-like syndrome	e71541	71	early infantile epileptic encephalopathy	221746	100	Early infantile epileptic encephalopathy	62653623 62641235 61749700 61749704 61753251
zellweger syndrome	e71510	67	usher syndrome, type 1b	197596	98	Usher syndrome, type 1	146733615 121908140 63749888 61750420 17847577 55843567 55843567 2234584 28934610 28934610 41298133 41298133 41298135 41298135 111033201

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