

## Additional File 5

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### **Common traits and mapping to ontologies**

Table 1: Traits from FinnGen and GWAS Catalog mapped to the same terms of EFO or NCIT. A few entries present ontology ids from other ontologies, but these are also contained in either EFO or NCIT.

| Ontology ID   | List of traits mapped to the ID from the two sources                                                                                                                                                                                                                                                  |
|---------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| EFO_0000274   | finngen: atopic dermatitis, gwas: atopic eczema, gwas: atopic eczema, covid-19                                                                                                                                                                                                                        |
| EFO_0000712   | finngen: accidents, finngen: stroke, finngen: stroke, excluding sah, finngen: stroke, including sah, gwas: stroke                                                                                                                                                                                     |
| EFO_0000729   | finngen: other ulcerative colitis, gwas: ulcerative colitis, crohn's disease                                                                                                                                                                                                                          |
| EFO_0001062   | finngen: cytomegaloviral disease, gwas: schizophrenia, cytomegalovirus infection                                                                                                                                                                                                                      |
| EFO_0003819   | finngen: dental caries, gwas: dental caries, gwas: dental caries, dentures                                                                                                                                                                                                                            |
| EFO_0003948   | finngen: gastro-oesophageal reflux disease, gwas: esophageal adenocarcinoma, gastroesophageal reflux disease                                                                                                                                                                                          |
| EFO_0004191   | finngen: androgenic alopecia, gwas: androgenetic alopecia                                                                                                                                                                                                                                             |
| EFO_0004257   | finngen: other neurotic disorders, gwas: neurotic disorder                                                                                                                                                                                                                                            |
| EFO_0004265   | finngen: peripheral artery disease, gwas: peripheral arterial disease                                                                                                                                                                                                                                 |
| EFO_0004683   | finngen: wet age-related macular degeneration, gwas: wet macular degeneration                                                                                                                                                                                                                         |
| EFO_0004772   | finngen: early onset copd, gwas: early onset hypertension                                                                                                                                                                                                                                             |
| EFO_0006812   | finngen: autoimmune thyroiditis, gwas: autoimmune thyroid disease                                                                                                                                                                                                                                     |
| EFO_0007135   | finngen: adult-onset still disease, gwas: adult-onset still's disease                                                                                                                                                                                                                                 |
| EFO_0009458   | finngen: alcohol use disorder, icd-based, finngen: alcohol use disorder, swedish definition, gwas: alcohol use disorder measurement, gwas: alcohol use disorder measurement, alcohol dependence                                                                                                       |
| EFO_0010822   | finngen: trigger finger, gwas: stenosing tenosynovitis                                                                                                                                                                                                                                                |
| EFO_1000786   | finngen: coxarthrosis, gwas: osteoarthritis, hip, gwas: osteoarthritis, hip, osteoarthritis, knee                                                                                                                                                                                                     |
| EFO_1000941   | finngen: adhesive capsulitis of shoulder, gwas: frozen shoulder                                                                                                                                                                                                                                       |
| EFO_1000965   | finngen: allergic purpura, gwas: henoch-schoenlein purpura                                                                                                                                                                                                                                            |
| EFO_1001178   | finngen: impingement syndrome of shoulder, gwas: shoulder impingement syndrome                                                                                                                                                                                                                        |
| EFO_1001250   | finngen: rotator cuff syndrome, gwas: rotator cuff tear                                                                                                                                                                                                                                               |
| EFO_1001436   | finngen: nontoxic single thyroid nodule, gwas: thyroid nodule                                                                                                                                                                                                                                         |
| EFO_1001494   | finngen: psoriasis vulgaris, finngen: psoriasis (vulgaris), strict definition, gwas: psoriasis vulgaris                                                                                                                                                                                               |
| EFO_1001506   | finngen: primary angle-closure glaucoma, gwas: primary angle closure glaucoma                                                                                                                                                                                                                         |
| EFO_1001507   | finngen: acohol-induced acute pancreatitis, gwas: asparaginase-induced acute pancreatitis                                                                                                                                                                                                             |
| HP_0000708    | finngen: behavioural disorders, gwas: behavioral abnormality                                                                                                                                                                                                                                          |
| HP_0100806    | finngen: puerperal sepsis, gwas: sepsis                                                                                                                                                                                                                                                               |
| MONDO_0005405 | finngen: childhood asthma (age16), gwas: childhood onset asthma                                                                                                                                                                                                                                       |
| NCIT.C114357  | finngen: other juvenile arthritis, gwas: juvenile idiopathic arthritis                                                                                                                                                                                                                                |
| NCIT.C117004  | finngen: migraine without aura, finngen: migraine without aura, drug-induced, gwas: migraine without aura                                                                                                                                                                                             |
| NCIT.C117005  | finngen: migraine with aura, finngen: migraine with aura, drug-induced, gwas: migraine with aura                                                                                                                                                                                                      |
| NCIT.C118764  | finngen: amblyopia ex anopsia, gwas: amblyopia                                                                                                                                                                                                                                                        |
| NCIT.C122822  | finngen: cholelithiasis, gwas: cholelithiasis, gallstones                                                                                                                                                                                                                                             |
| NCIT.C142891  | finngen: cr(e)st syndrome, gwas: brugada syndrome                                                                                                                                                                                                                                                     |
| NCIT.C157552  | finngen: hypertension, pulmonary arterial, gwas: pulmonary arterial hypertension                                                                                                                                                                                                                      |
| NCIT.C164315  | finngen: interstitial lung disease, finngen: interstitial lung disease endpoints, gwas: interstitial lung disease                                                                                                                                                                                     |
| NCIT.C26696   | finngen: all anxiety disorders, finngen: phobic anxiety disorders, gwas: anxiety                                                                                                                                                                                                                      |
| NCIT.C26713   | finngen: other cataract, gwas: cataract                                                                                                                                                                                                                                                               |
| NCIT.C26748   | finngen: diabetic neuropathy, gwas: diabetic foot, neuropathy, gwas: diabetic neuropathy                                                                                                                                                                                                              |
| NCIT.C26782   | finngen: glaucoma, finngen: glaucoma suspect, finngen: normotensive glaucoma, gwas: glaucoma                                                                                                                                                                                                          |
| NCIT.C26800   | finngen: hypothyroidism, levothyroxin purchases, finngen: hypothyroidism, other/unspecified, finngen: hypothyroidism, strict autoimmune, finngen: postinfectious hypothyroidism, gwas: hypothyroidism                                                                                                 |
| NCIT.C26845   | finngen: parkinson's disease, finngen: parkinson's disease, including avohilmo, finngen: parkinson's disease, strict definition, gwas: parkinson's disease                                                                                                                                            |
| NCIT.C27195   | finngen: acne, finngen: acne vulgaris, gwas: acne                                                                                                                                                                                                                                                     |
| NCIT.C27996   | finngen: complications following myocardial infarction, finngen: myocardial infarction, finngen: myocardial infarction, strict, gwas: myocardial infarction                                                                                                                                           |
| NCIT.C28397   | finngen: asthma, finngen: asthma, including avohilmo, gwas: allergy, asthma, gwas: asthma                                                                                                                                                                                                             |
| NCIT.C2884    | finngen: other/unspecified rheumatoid arthritis, finngen: rheumatoid arthritis, finngen: seronegative rheumatoid arthritis, finngen: seropositive rheumatoid arthritis, finngen: seropositive rheumatoid arthritis, wide, gwas: rheumatoid arthritis, gwas: rheumatoid arthritis, follicular lymphoma |
| NCIT.C2952    | finngen: ulcerative colitis, finngen: ulcerative colitis, nas, gwas: rheumatoid arthritis, ulcerative colitis, gwas: ulcerative colitis                                                                                                                                                               |
| NCIT.C2985    | finngen: diabetes mellitus, finngen: diabetes, varying definitions, gwas: diabetes mellitus                                                                                                                                                                                                           |
| NCIT.C3001    | finngen: dermatitis and eczema, gwas: eczema                                                                                                                                                                                                                                                          |
| NCIT.C3123    | finngen: autoimmune hyperthyroidism, gwas: hyperthyroidism                                                                                                                                                                                                                                            |
| NCIT.C3163    | finngen: chronic lymphocytic leukaemia, gwas: chronic lymphocytic leukemia, gwas: multiple sclerosis, chronic lymphocytic leukemia, gwas: rheumatoid arthritis, chronic lymphocytic leukemia                                                                                                          |
| NCIT.C3199    | finngen: copd, finngen: other chronic obstructive pulmonary disease, gwas: chronic obstructive pulmonary disease, gwas: chronic obstructive pulmonary disease, asthma, gwas: hypertension, chronic obstructive pulmonary disease                                                                      |
| NCIT.C3201    | finngen: systemic lupus erythematosus, unspecified, gwas: rheumatoid arthritis, systemic lupus erythematosus, gwas: systemic sclerosis, systemic lupus erythematosus                                                                                                                                  |
| NCIT.C3209    | finngen: follicular lymphoma, finngen: non-follicular lymphoma, gwas: multiple sclerosis, follicular lymphoma                                                                                                                                                                                         |
| NCIT.C3243    | finngen: ms-disease / multiple sclerosis, gwas: multiple sclerosis, gwas: multiple sclerosis, disease recurrence, gwas: multiple sclerosis, triglyceride measurement                                                                                                                                  |
| NCIT.C3256    | finngen: nasal polyp, gwas: nasal cavity polyp                                                                                                                                                                                                                                                        |
| NCIT.C3298    | finngen: osteoporosis, gwas: obesity, osteoporosis, gwas: osteoporosis                                                                                                                                                                                                                                |
| NCIT.C3303    | finngen: pain in joint, finngen: tmd related pain, gwas: pain                                                                                                                                                                                                                                         |
| NCIT.C3423    | finngen: respiratory tuberculosis, finngen: respiratory tuberculosis, finngen: tuberculosis, gwas: tuberculosis                                                                                                                                                                                       |
| NCIT.C3444    | finngen: wegner granulomatosis, gwas: granulomatosis with polyangiitis                                                                                                                                                                                                                                |
| NCIT.C34440   | finngen: bulimia nervosa (incl. atypical), gwas: bulimia nervosa                                                                                                                                                                                                                                      |
| NCIT.C34525   | finngen: vascular dementia, finngen: vascular dementia (mixed), finngen: vascular dementia (undefined), gwas: vascular dementia                                                                                                                                                                       |
| NCIT.C34538   | finngen: diabetic retinopathy, finngen: other diabetic retinopathy, finngen: unspecified diabetic retinopathy, gwas: diabetic retinopathy                                                                                                                                                             |
| NCIT.C34549   | finngen: disturbances of skin sensation, gwas: disturbance of skin sensation                                                                                                                                                                                                                          |
| NCIT.C34650   | finngen: gout, finngen: gout, finngen, finngen: gout, strict definition, finngen: gout, unspecified, finngen: idiopathic gout, gwas: gout                                                                                                                                                             |
| NCIT.C34658   | finngen: hallux valgus (acquired), gwas: hallux valgus                                                                                                                                                                                                                                                |
| NCIT.C34690   | finngen: inguinal hernia, finngen: inguinal hernia, bilateral, finngen: inguinal hernia, unilateral, gwas: inguinal hernia                                                                                                                                                                            |
| NCIT.C34882   | finngen: rheumatic fever incl heart disease, gwas: rheumatic heart disease                                                                                                                                                                                                                            |
| NCIT.C34885   | finngen: otitis media, unspecified, gwas: otitis media                                                                                                                                                                                                                                                |
| NCIT.C34890   | finngen: panic disorder, gwas: anxiety disorder, panic disorder, gwas: panic disorder                                                                                                                                                                                                                 |
| NCIT.C34942   | finngen: gestational diabetes (for exclusion), gwas: gestational diabetes                                                                                                                                                                                                                             |
| NCIT.C35040   | finngen: other strabismus, finngen: strabismus, finngen: vertical strabismus, gwas: strabismus                                                                                                                                                                                                        |
| NCIT.C35065   | finngen: giant cell arteritis, finngen: temporal arteritis, gwas: temporal arteritis                                                                                                                                                                                                                  |
| NCIT.C3510    | finngen: malignant melanoma of skin, gwas: cutaneous melanoma, gwas: neuroblastoma, cutaneous melanoma                                                                                                                                                                                                |
| NCIT.C35145   | finngen: acute appendicitis, finngen: appendicitis, broad definition, finngen: other appendicitis, gwas: appendicitis                                                                                                                                                                                 |
| NCIT.C35731   | finngen: other hearing loss, gwas: hearing loss                                                                                                                                                                                                                                                       |
| NCIT.C35768   | finngen: peripheral atherosclerosis, gwas: atherosclerosis                                                                                                                                                                                                                                            |
| NCIT.C38012   | finngen: isolated proteinuria, gwas: proteinuria                                                                                                                                                                                                                                                      |
| NCIT.C4731    | finngen: diabetich neuropathy, finngen: intercostal neuropathy, gwas: neuropathy                                                                                                                                                                                                                      |
| NCIT.C50577   | finngen: all-cause heart failure, finngen: heart failure, not strict, finngen: heart failure,strict, gwas: heart failure                                                                                                                                                                              |
| NCIT.C61277   | finngen: psoriatic arthropathies, gwas: psoriatic arthritis                                                                                                                                                                                                                                           |
| NCIT.C63709   | finngen: temporomandibular joint disorders, gwas: temporomandibular joint disorder                                                                                                                                                                                                                    |
| NCIT.C72070   | finngen: systemic sclerosis, gwas: systemic scleroderma, gwas: systemic scleroderma, crohn's disease                                                                                                                                                                                                  |
| NCIT.C79532   | finngen: allergic rhinitis, gwas: allergic rhinitis, gwas: eczema, allergic rhinitis, gwas: eczema, allergic rhinitis, asthma                                                                                                                                                                         |
| NCIT.C80078   | finngen: chronic kidney disease, gwas: chronic kidney disease, gwas: chronic kidney disease, diabetic nephropathy, gwas: creatinine measurement, chronic kidney disease                                                                                                                               |
| NCIT.C84417   | finngen: diabetic nephropathy, gwas: albuminuria, diabetic nephropathy, gwas: diabetic nephropathy                                                                                                                                                                                                    |
| NCIT.C84564   | finngen: ankylosing spondylitis, finngen: ankylosing spondylitis, strict definition, gwas: ankylosing spondylitis                                                                                                                                                                                     |
| NCIT.C8851    | finngen: diffuse large b-cell lymphoma, gwas: diffuse large b-cell lymphoma, gwas: diffuse large b-cell lymphoma, rheumatoid arthritis                                                                                                                                                                |
| NCIT.C94378   | finngen: schizoaffective disorder, gwas: schizophrenia, schizoaffective disorder                                                                                                                                                                                                                      |
| NCIT.C94569   | finngen: tooth eruption problems, gwas: tooth eruption                                                                                                                                                                                                                                                |
| NCIT.C97161   | finngen: autism, finngen: autism spe, gwas: autism                                                                                                                                                                                                                                                    |