

Supplementary information 1.3- HPO terms

HPO ID	Clinical Feature	Present	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID
HP:0000010	Recurrent UTIs	Yes	001	008	009	013	014											
HP:0000024	Prostatitis	Yes	006															
HP:0000050	Hypoplastic genitalia	Yes	015	016	017													
HP:0000076	Vesicoureteral reflux	Yes	001															
HP:0000103	Polyuria	Yes	005	006	015													
HP:0000107	Renal cyst	Yes	003	005	018													
HP:0000131	Uterine leiomyoma	Yes	005	014														
HP:0000132	Menorrhagia	Yes	013	014														
HP:0000135	Hypogonadism	Yes	006	016	017													
HP:0000147	Polycystic ovaries	Yes	013	014														
HP:0000218	High palate	Yes	001	006	007	008	010	012	018									
HP:0000272	Malar flattening	Yes	008															
HP:0000403	Recurrent otitis media	Yes	013															
HP:0000486	Strabismus	Yes	007	008														
HP:0000498	Blepharitis	Yes	001	004	005	006	008	009	011	012	014							
HP:0000510	Rod-cone dystrophy	Yes	001	002	003	004	005	006	007	008	009	010	011	012	013	014	015	018
HP:0000510	Rod-cone dystrophy	No	016	017			010											
HP:0000518	Cataract	Yes	001	005	006	008		011	012									
HP:0000618	Blindness	Yes	001	002	004	005	006	009	010	011	012	013	014	018				
HP:0000639	Nystagmus	Yes	005															
HP:0000662	Nyctalopia	Yes	001	002	005	009	015											
HP:0000678	Dental crowding	Yes	005	006	007	008	010	012	018									
HP:0000716	Depression	Yes	003	006	014													
HP:0000750	Delayed speech	Yes	002	007	010	016	017	018										
HP:0000787	Nephrolithiasis	Yes	005	012														
HP:0000821	Hypothyroidism	Yes	003	007														
HP:0000822	Hypertension	Yes	002	003	005													
HP:0000873	Diabetes insipidus	Yes	013															
HP:0000975	Hyperhidrosis	Yes	006															
HP:0001007	Hirsutism	Yes	001															
HP:0001061	Acne	Yes	013															
HP:0001128	Trichiasis	Yes	004															
HP:0001156	Brachydactyly	Yes	002	006	013	014	018											
HP:0001161	Postaxial hand polydactyly	Yes	001	003	006	010	013	014	016	017	018							

Table A2.1: Human Phenotype Ontology (HPO) terms for Bardet Biedl syndrome (BBS) cohort

HPO ID	Clinical Feature	Present	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID
HP:0001182	Tapered finger	Yes	001															
HP:0001249	Intellectual disability	Yes	002	003	005	006	007	008	009	010	011	012	013	014	015	016	017	
HP:0001249	Intellectual disability	No	001	004	018													
HP:0001252	Muscular hypotonia	Yes	006	018														
HP:0001256	Intellectual disability, mild	Yes																
HP:0001281	Tetany	Yes	014															
HP:0001369	Arthritis	Yes	011	012														
HP:0001407	Hepatic cysts	Yes	018															
HP:0001513	Obesity	Yes	001	002	003	005	006	007	008	010	011	012	014	016	017			
HP:0001513	Obesity	No	004	009	013	018												
HP:0001622	Premature birth	Yes	013															
HP:0001650	Aortic valve stenosis	Yes	013															
HP:0001681	Angina pectoris	Yes	005															
HP:0001696	Situs inversus totalis	Yes	018															
HP:0001748	Polysplenia	Yes	013															
HP:0001829	Postaxial foot polydactyly	Yes	002	005	007	008	009	014	018									
HP:0001831	Short toe	Yes	001	005														
HP:0001852	Sandal gap	Yes	001															
HP:0001959	Polydipsia	Yes	006	015														
HP:0001997	Gout	Yes	015															
HP:0002360	Sleep disturbance	Yes	012	014														
HP:0002370	Poor coordination	Yes	015	018														
HP:0002566	Intestinal malrotation	Yes	013															
HP:0002608	Celiac disease	Yes	009															
HP:0002619	Varicose veins	Yes	010	011	013													
HP:0002870	Obstructive sleep apnoea	Yes	008	009														
HP:0003124	Hypercholesterolemia	Yes	006															
HP:0003225	Reduced factor V activity	Yes	001															
HP:0003326	Myalgia	Yes	006															
HP:0003416	Spinal canal stenosis	Yes	002															
HP:0003764	Nevus	Yes	005															
HP:0003765	Psoriasis	Yes	006															
HP:0003774	Stage 5 CKD	Yes	014	018														
HP:0004322	Short stature	Yes	001	002	003	004	006	007	008	009	010	011	012	013	014	015	018	
HP:0004322	Short stature	No	005	016	017													

Table A2.1 continued: Human Phenotype Ontology (HPO) terms for Bardet Biedl syndrome (BBS) cohort

HPO ID	Clinical Feature	Present	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID
HP:0004936	Venous thrombosis	Yes	012															
HP:0005059	Arthralgia/arthritis	Yes	013															
HP:0005263	Gastritis	Yes	013															
HP:0005978	Type II diabetes mellitus	Yes	002															
HP:0007133	Peripheral neuropathy	Yes	002															
HP:0007302	Bipolar affective disorder	Yes	006	007														
HP:0007401	Macular atrophy	Yes	001															
HP:0010442	Polydactyly	No	001	004	011	012	015											
HP:0010522	Dyslexia	Yes	009															
HP:0010605	Chalazion	Yes	008															
HP:0010677	Enuresis nocturna	Yes	006															
HP:0011671	Interrupted vena cava	Yes	013															
HP:0011800	Midface retrusion	Yes	007															
HP:0012330	Pyelonephritis	Yes	001	005														
HP:0012432	Chronic fatigue	Yes	013															
HP:0012450	Chronic constipation	Yes	013															
HP:0012540	Axillary epidermoid cyst	Yes	013															
HP:0012622	Chronic kidney disease	Yes	007	014														
HP:0100280	Crohn's disease	Yes	018															
HP:0100651	Type I diabetes mellitus	Yes	008															
HP:0100753	Schizophrenia	Yes	006	007														

Table A2.1 continued: Human Phenotype Ontology (HPO) terms for Bardet Biedl syndrome (BBS) cohort

HPO ID	Clinical Feature	Present	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID
HP:0000010	Recurrent UTIs	Yes	016													
HP:0000093	Proteinuria	Yes	008													
HP:0000143	Rectovaginal fistula	Yes	017													
HP:0000230	Gingivitis	Yes	014													
HP:0000252	Microcephaly	Yes	009													
HP:0000407	Sensorineural hearing loss	Yes	017	021												
HP:0000501	Glaucoma	Yes	008													
HP:0000526	Aniridia	Yes	008													
HP:0000540	Hypermetropia	Yes	021													
HP:0000707	Abnormality of CNS	Yes	010													
HP:0000717	Autism	Yes	020													
HP:0000735	Impaired social interactions	Yes	020													
HP:0000750	Delayed speech	Yes	008	020												
HP:0000822	Hypertension	Yes	017													
HP:0000824	Growth hormone deficiency	Yes	016													
HP:0000846	Adrenal insufficiency	Yes	011	015												
HP:0000951	Abnormality of the skin	Yes	019													
HP:0000964	Eczema	Yes	001	005	006	010	019									
HP:0000988	Skin rash	Yes	012													
HP:0001047	Atopic dermatitis	Yes	011	018												
HP:0001081	Cholelithiasis	Yes	021													
HP:0001250	Seizures	Yes	017	019												
HP:0001251	Ataxia	Yes	019													
HP:0001254	Lethargy	Yes	011													
HP:0001257	Spasticity	Yes	019													
HP:0001263	Developmental delay	Yes	009	016	017											
HP:0001287	Meningitis	Yes	016													
HP:0001332	Dystonia	Yes	019													

Table A2.2 Human Phenotype Ontology (HPO) terms for very early onset inflammatory bowel disease (IBD) cohort

HPO ID	Clinical Feature	Present	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID
HP:0001382	Joint hypermobility	Yes	010													
HP:0001510	Growth delay	Yes	008	017	019	020										
HP:0001525	Severe failure to thrive	Yes	002													
HP:0001562	Oligohydramnios	Yes	008													
HP:0001622	Premature birth	Yes	008	009	011	021										
HP:0001629	Ventricular septal defect	Yes	012													
HP:0001655	Patent foramen ovale	Yes	008													
HP:0001662	Bradycardia	Yes	016													
HP:0001684	Atrial septal defect	Yes	016													
HP:0011410	Caesarean section	Yes	003	007	008	010	013	021								
HP:0001875	Neutropenia	Yes	016													
HP:0001876	Pancytopenia	Yes	021													
HP:0001880	Eosinophilia	Yes	010	016												
HP:0001891	Iron deficiency anaemia	Yes	017	004	011	015										
HP:0001943	Hypoglycaemia	Yes	006	016												
HP:0001999	Abnormal facial shape	Yes	008	009												
HP:0002011	CNS malformation	Yes	019													
HP:0002013	Vomiting	Yes	010	015	016	017										
HP:0002014	Diarrhoea	Yes	003	005	006	009	012	013	015	016	017	018	019	020		
HP:0002019	Constipation	Yes	002	013	014	016	020									
HP:0002020	Gastroesophageal reflux	Yes	011													
HP:0002027	Abdominal pain	Yes	001	002	003	005	006	010	011	012	013	014	016	017	019	020
HP:0002034	Abnormality of rectum	Yes	009													
HP:0002037	Inflammation of large intestine	Yes	001	003	005	006	007	008	013	015	020	019	021			
HP:0002043	Oesophageal stricture	Yes	020													
HP:0002072	Chorea	Yes	019													
HP:0002098	Respiratory distress	Yes	021													
HP:0002240	Hepatomegaly	Yes	016													

Table A2.2 continued: Human Phenotype Ontology (HPO) terms for very early onset inflammatory bowel disease (IBD) cohort

HPO ID	Clinical Feature	Present	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID
HP:0002315	Headache	Yes	016													
HP:0002373	Febrile seizures	Yes	012	015												
HP:0002500	White matter abnormality	Yes	021													
HP:0002566	Intestinal malrotation	Yes	009													
HP:0002573	Haematochezia	Yes	003	004	007	010	011	012	013	014	015	017	019	020		
HP:0002579	Gastrointestinal dysmotility	Yes	010													
HP:0002583	Colitis	Yes	004	007	013	016	017	018	019							
HP:0002589	Gastrointestinal atresia	Yes	021													
HP:0002607	Bowel incontinence	Yes	009													
HP:0002633	Vasculitis	Yes	012	013												
HP:0002719	Recurrent infections	Yes	021													
HP:0002750	Delayed skeletal maturation	Yes	001	003												
HP:0002788	Recurrent RTIs	Yes	005	008												
HP:0002870	Obstructive sleep apnoea	Yes	008													
HP:0002955	Granulomatosis	Yes	002	018												
HP:0002960	Autoimmunity	Yes	017													
HP:0003765	Psoriasis	Yes	019													
HP:0004322	Short stature	Yes	003	001	008	010	011	014	015	016	017	018				
HP:0004325	Decreased body weight	Yes	015	017	018											
HP:0004349	Reduced bone density	Yes	001	017												
HP:0004387	Enterocolitis	Yes	021													
HP:0004399	Congenital pyloric atresia	Yes	021													
HP:0004797	Small bowel atresias	Yes	009													
HP:0006568	Increased hepatic glycogen	Yes	016													
HP:0006846	Acute encephalopathy	Yes	017													
HP:0007018	ADHD	Yes	010													
HP:0007359	Focal seizures	Yes	016													
HP:0007385	Aplasia cutis congenita	Yes	011													

Table A2.2 continued: Human Phenotype Ontology (HPO) terms for very early onset inflammatory bowel disease (IBD) cohort

HPO ID	Clinical Feature	Present	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID
HP:0008169	Reduced factor VII activity	Yes	016													
HP:0009789	Perianal abscess	Yes	005	017												
HP:0010301	Spinal dysraphism	Yes	019													
HP:0010447	Anal fistula	Yes	005													
HP:0010609	Skin tags	Yes	014	018												
HP:0011100	Intestinal atresia	Yes	021													
HP:0011102	Ileal atresia	Yes	021													
HP:0011107	Aphthous stomatitis	Yes	002	014												
HP:0011473	Villous atrophy	Yes	005	010	021											
HP:0011858	Reduced factor IX activity	Yes	016													
HP:0011900	Hyperfibrinogenaemia	Yes	018													
HP:0012390	Anal fissure	Yes	002	014												
HP:0012390	Anal fissure	Yes	014													
HP:0012537	Food intolerance	Yes	004													
HP:0030021	Auricular tag	Yes	011													
HP:0030318	Angular cheilitis	Yes	014	018												
HP:0100279	Ulcerative colitis	Yes	006	019	002											
HP:0100280	Crohn's disease	Yes	002	005	013	014	018	020								
HP:0100633	Esophagitis	Yes	001													
HP:0100806	Sepsis	Yes	016													
HP:0100825	Cheilitis	Yes	018													
HP:0100838	Recurrent skin abscess	Yes	009													
Table A2.2 continued: Human Phenotype Ontology (HPO) terms for very early onset inflammatory bowel disease (IBD) cohort																

HPO ID	Clinical Feature	Present	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID
HP:0000020	Urinary incontinence	Yes													
HP:0000020	Urinary incontinence	No	001	002	003	004	005	006	007	008	009	010	011	012	013
HP:0000103	Polyuria	Yes													
HP:0000103	Polyuria	No	001	002	003	004	005	006	007	008	009	010	011	012	013
HP:0000155	Oral ulcer	Yes	007	008	011										
HP:0000155	Oral ulcer	No	001	002	003	004	005	006	009	010	012	013			
HP:0000969	Oedema	Yes	002	009	010	011	012								
HP:0000969	Oedema	No	001	003	004	005	006	007	008	013					
HP:0000988	Skin rash	Yes	006	007	008	010	011	012							
HP:0000988	Skin rash	No	001	002	003	004	005	009	013						
HP:0001369	Arthritis	Yes	003	007	011										
HP:0001369	Arthritis	No	001	002	004	005	006	008	009	010	012	013			
HP:0001386	Joint swelling	Yes	003	007	011										
HP:0001386	Joint swelling	No	001	002	004	005	006	008	009	010	012	013			
HP:0001387	Joint stiffness	Yes	002	003	007	011									
HP:0001387	Joint stiffness	No	001	004	005	006	008	009	010	012	013				
HP:0001596	Alopecia	Yes	011												
HP:0001596	Alopecia	No	001	002	003	004	005	006	007	008	009	010	012	013	
HP:0001597	Nail fold changes	Yes	001	002	004	007	008	009	010	012	013				
HP:0001597	Nail fold changes	No	003	005	006	011									
HP:0001618	Dysphonia	Yes	002	004											
HP:0001618	Dysphonia	No	001	003	005	006	007	008	009	010	011	012	013		
HP:0001824	Weight loss	Yes	007	010	011										
HP:0001824	Weight loss	No	001	002	003	004	005	006	008	009	012	013			
HP:0001945	Fever	Yes	001	009											
HP:0001945	Fever	No	002	003	004	005	006	007	008	010	011	012	013		
HP:0002014	Diarrhoea	Yes	001												
HP:0002014	Diarrhoea	No	002	003	004	005	006	007	008	009	010	011	012	013	
HP:0002027	Abdominal pain	Yes	001	011											
HP:0002027	Abdominal pain	No	002	003	004	005	006	007	008	009	010	012	013		
HP:0002094	Dyspnoea	Yes													
HP:0002094	Dyspnoea	No	001	002	003	004	005	006	007	008	009	010	011	012	013
HP:0002249	Melaena	Yes													

Table A2.3: Human Phenotype Ontology (HPO) terms for juvenile dermatomyositis (JDM) cohort

HPO ID	Clinical Feature	Present	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID	ID
HP:0002249	Melaena	No	001	002	003	004	005	006	007	008	009	010	011	012	013
HP:0002315	Headache	Yes	003	004	007	008	011	013							
HP:0002315	Headache	No	001	003	005	006	009	010	012						
HP:0002829	Arthralgia	Yes	001	002	003	006	007	009	010	012	013				
HP:0002829	Arthralgia	No	004	005	008	011									
HP:0002960	Autoimmunity	Yes	009												
HP:0002960	Autoimmunity	No	001	002	003	004	005	006	007	008	010	011	012	013	
HP:0003324	Muscle weakness	Yes	001	002	003	004	005	006	007	008	009	010	011	012	013
HP:0003324	Muscle weakness	No													
HP:0003326	Myalgia	Yes	001	002	003	002	002	006	007	008	009	010	011	012	013
HP:0003326	Myalgia	No													
HP:0003761	Calcinosis	Yes	001												
HP:0003761	Calcinosis	No	002	003	004	005	006	007	008	009	010	011	012	013	
HP:0011107	Aphthous stomatitis	Yes													
HP:0011107	Aphthous stomatitis	No	001	002	003	004	005	006	007	008	009	010	011	012	013
HP:0012369	Malar (heliotrope) rash	Yes	006	012											
HP:0012369	Malar (heliotrope) rash	No	001	002	003	004	005	007	008	009	010	011	013		
HP:0012378	Fatigue	Yes	001	002	003	004	005	006	007	008	009	010	011	012	013
HP:0012378	Fatigue	No													
HP:0100578	Lipoatrophy	Yes													
HP:0100578	Lipoatrophy	No	001	002	003	004	005	006	007	008	009	010	011	012	013
HP:0100749	Chest pain	Yes													
HP:0100749	Chest pain	No	001	002	003	004	005	006	007	008	009	010	011	012	013
HP:0200034	Papule	Yes	007	011											
HP:0200034	Papule	No	001	002	003	004	005	006	008	009	010	012	013		
HP:0200042	Skin ulcer	Yes	001	003	012										
HP:0200042	Skin ulcer	No	002	004	005	006	007	008	009	010	011	013			
Not in HPO	Gotttron's papules	Yes	002	005	006	007	008	009	011						
Not in HPO	Gotttron's papules	No	001	003	004	010	012	013							
Not in HPO	History of trauma	Yes	002												
Not in HPO	History of trauma	No	001	003	004	005	006	007	008	009	010	011	012	013	
Not in HPO	Raynaud's phenomenon	Yes	001												
Not in HPO	Raynaud's phenomenon	No	002	003	004	005	006	007	008	009	010	011	012	013	

Table A2.3 continued: Human Phenotype Ontology (HPO) terms for juvenile dermatomyositis (JDM) cohort

HPO ID	Clinical Feature	Present	ID	ID
HP:0000510	Rod-cone dystrophy	Yes	001	002
HP:0000618	Blindness	Yes	001	002
HP:0001382	Joint hypermobility	Yes	001	
HP:0001751	Vestibular dysfunction	Yes	002	
HP:0001751	Vestibular dysfunction	No	001	
HP:0002076	Migraine	Yes	002	
HP:0003418	Back pain	Yes	001	
HP:0008527	Congenital sensorineural hearing loss	Yes	001	002
HP:0012173	Orthostatic tachycardia	Yes	001	
Table A2.4: Human Phenotype Ontology (HPO) terms for Usher syndrome (USH) cohort				