Lab notebook

KBC

4/5/2021

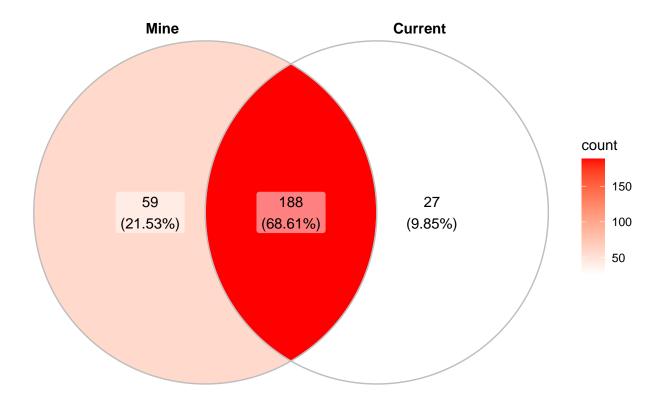
2021-04-05 report

Utility functions

Opposites already manually paired

Bill can explain where these came from better than I can.

```
## Warning: package 'ggVennDiagram' was built under R version 4.0.2
## Warning: package 'tidyverse' was built under R version 4.0.2
## -- Attaching packages ------ tidyverse
## v ggplot2 3.3.2
                 v purrr 0.3.4
## v tibble 3.0.1
                 v dplyr 1.0.4
                v stringr 1.4.0
## v tidyr 1.1.1
## v readr 1.4.0
                 v forcats 0.5.1
## Warning: package 'ggplot2' was built under R version 4.0.2
## Warning: package 'tidyr' was built under R version 4.0.2
## Warning: package 'readr' was built under R version 4.0.2
## Warning: package 'dplyr' was built under R version 4.0.2
## Warning: package 'forcats' was built under R version 4.0.2
## -- Conflicts ------ tidyverse_confl
## x dplyr::filter() masks stats::filter()
## x dplyr::lag() masks stats::lag()
```



```
## [1] "IN BOTH MINE AND THE CURRENT PAIRS:"
```

- ## [1] "abnormal" "normal"
- ## [1] "absent" "present"
- ## [1] "active" "inactive"
- ## [1] "acute" "chronic"
- ## [1] "adhesive" "non-adhesive"
- ## [1] "aerobic" "anaerobic"
- ## [1] "aerobic (for occurrence)" "anaerobic (for occurrence)"
- ## [1] "aligned with" "misaligned with"
- ## [1] "aromatic" "non-aromatic"
- ## [1] "arrhythmic" "rhythmic"
- ## [1] "asynchronous" "synchronous"
- ## [1] "back" "front"
- ## [1] "balanced" "unbalanced"
- ## [1] "branched" "unbranched"
- ## [1] "closed" "open"
- ## [1] "coiled" "uncoiled"
- ## [1] "colored" "discolored"
- ## [1] "compatible" "incompatible"
- ## [1] "complete structure" "incomplete structure"
- ## [1] "condensed" "decondensed"
- ## [1] "conspicuous" "inconspicuous"
- ## [1] "continuous" "discontinuous"
- ## [1] "contractile" "non-contractile"

```
## [1] "coordinated"
                       "uncoordinated"
## [1] "crowded"
                   "uncrowded"
## [1] "damaged"
                   "undamaged"
## [1] "decreased accumulation" "increased accumulation"
## [1] "decreased acidity" "increased acidity"
## [1] "decreased adhesivity" "increased adhesivity"
## [1] "decreased affinity" "increased affinity"
## [1] "decreased age" "increased age"
## [1] "decreased amount" "increased amount"
## [1] "decreased angle to" "increased angle to"
## [1] "decreased anterior-posterior diameter"
## [2] "increased anterior-posterior diameter"
## [1] "decreased area" "increased area"
## [1] "decreased avidity" "increased avidity"
## [1] "decreased behavioural activity" "increased behavioural activity"
## [1] "decreased branchiness" "increased branchiness"
## [1] "decreased cellular motility" "increased cellular motility"
## [1] "decreased circumference" "increased circumference"
## [1] "decreased coiling" "increased coiling"
## [1] "decreased combustibility" "increased combustibility"
## [1] "decreased concentration" "increased concentration"
## [1] "decreased contractility" "increased contractility"
## [1] "decreased coordination" "increased coordination"
## [1] "decreased curvature" "increased curvature"
## [1] "decreased degree of illumination" "increased degree of illumination"
## [1] "decreased depth" "increased depth"
## [1] "decreased diameter" "increased diameter"
## [1] "decreased distance" "increased distance"
## [1] "decreased distribution" "increased distribution"
## [1] "decreased duration" "increased duration"
## [1] "decreased duration of temperature" "increased duration of temperature"
## [1] "decreased efficacy" "increased efficacy"
## [1] "decreased efficiency" "increased efficiency"
## [1] "decreased elasticity" "increased elasticity"
## [1] "decreased elevation" "increased elevation"
## [1] "decreased fatigability" "increased fatigability"
## [1] "decreased fecundity" "increased fecundity"
## [1] "decreased female fertility" "increased female fertility"
## [1] "decreased female receptivity" "increased female receptivity"
## [1] "decreased fertility" "increased fertility"
## [1] "decreased flexibility" "increased flexibility"
## [1] "decreased fluid flow" "increased fluid flow"
## [1] "decreased fluorescence" "increased fluorescence"
## [1] "decreased force" "increased force"
## [1] "decreased fragility" "increased fragility"
## [1] "decreased frequency" "increased frequency"
## [1] "decreased functionality" "increased functionality"
## [1] "decreased height" "increased height"
## [1] "decreased humidity" "increased humidity"
## [1] "decreased intensity" "increased intensity"
## [1] "decreased length" "increased length"
## [1] "decreased life span" "increased life span"
## [1] "decreased magnetism" "increased magnetism"
## [1] "decreased magnitude" "increased magnitude"
```

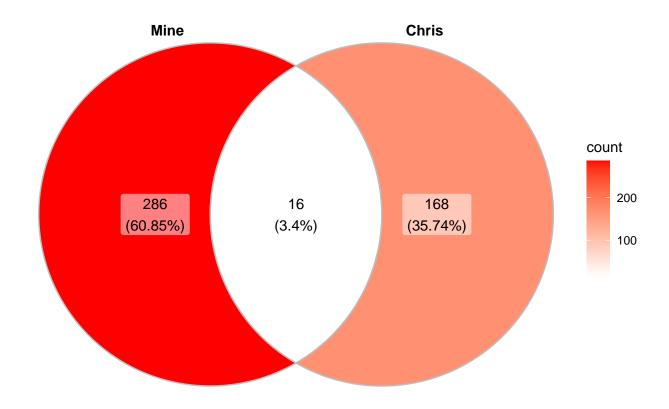
```
## [1] "decreased male fertility" "increased male fertility"
## [1] "decreased male receptivity" "increased male receptivity"
## [1] "decreased mass" "increased mass"
## [1] "decreased mass density" "increased mass density"
## [1] "decreased mobility" "increased mobility"
## [1] "decreased object quality" "increased object quality"
## [1] "decreased occurrence" "increased occurrence"
## [1] "decreased odor" "increased odor"
## [1] "decreased osmolality" "increased osmolality"
## [1] "decreased osmolarity" "increased osmolarity"
## [1] "decreased perimeter" "increased perimeter"
## [1] "decreased permeability" "increased permeability"
## [1] "decreased phosphorylation" "increased phosphorylation"
## [1] "decreased photosensitivity" "increased photosensitivity"
## [1] "decreased pigmentation" "increased pigmentation"
## [1] "decreased porosity" "increased porosity"
## [1] "decreased position" "increased position"
## [1] "decreased pressure" "increased pressure"
## [1] "decreased process quality" "increased process quality"
## [1] "decreased quality" "increased quality"
## [1] "decreased radioactivity" "increased radioactivity"
## [1] "decreased radiopacity" "increased radiopacity"
## [1] "decreased rate" "increased rate"
## [1] "decreased resistance to" "increased resistance to"
## [1] "decreased sensitivity of a process" "increased sensitivity of a process"
## [1] "decreased sensitivity of a process to oxygen"
## [2] "increased sensitivity of a process to oxygen"
## [1] "decreased sensitivity to irradiation"
## [2] "increased sensitivity to irradiation"
## [1] "decreased sensitivity toward" "increased sensitivity toward"
## [1] "decreased size" "increased size"
## [1] "decreased solubility" "increased solubility"
## [1] "decreased speed" "increased speed"
## [1] "decreased stability" "increased stability"
## [1] "decreased strength" "increased strength"
## [1] "decreased susceptibility toward" "increased susceptibility toward"
## [1] "decreased temperature" "increased temperature"
## [1] "decreased tendency" "increased tendency"
## [1] "decreased thickness" "increased thickness"
## [1] "decreased threshold" "increased threshold"
## [1] "decreased tolerance to" "increased tolerance to"
## [1] "decreased tonicity" "increased tonicity"
## [1] "decreased turgor" "increased turgor"
## [1] "decreased variability" "increased variability"
## [1] "decreased variability of color" "increased variability of color"
## [1] "decreased variability of rate" "increased variability of rate"
## [1] "decreased variability of size" "increased variability of size"
## [1] "decreased variability of temperature"
## [2] "increased variability of temperature"
## [1] "decreased velocity" "increased velocity"
## [1] "decreased viscosity" "increased viscosity"
## [1] "decreased volume" "increased volume"
## [1] "decreased waxiness" "increased waxiness"
## [1] "decreased weight" "increased weight"
```

```
## [1] "decreased wetness" "increased wetness"
## [1] "decreased width" "increased width"
## [1] "decreased width and length" "increased width and length"
## [1] "defasciculated" "fasciculated"
## [1] "degenerate"
                       "non-degenerate"
## [1] "dephosphorylated" "phosphorylated"
## [1] "differentiated" "undifferentiated"
                      "indiscriminate"
## [1] "discriminate"
## [1] "disorganized" "organized"
## [1] "disoriented" "oriented"
## [1] "distributed"
                       "undistributed"
## [1] "down" "up"
## [1] "dry" "wet"
## [1] "edible" "inedible"
## [1] "efficient" "inefficient"
                 "inelastic"
## [1] "elastic"
## [1] "elongated" "shortened"
## [1] "female fertile" "female sterile"
## [1] "female semi-fertile" "female semi-sterile"
## [1] "fertile" "sterile"
                   "inflexible"
## [1] "flexible"
## [1] "fragile"
                     "non-fragile"
## [1] "functional"
                        "non-functional"
## [1] "hard" "soft"
## [1] "high brightness" "low brightness"
## [1] "high saturation" "low saturation"
## [1] "immature" "mature"
## [1] "immobile" "mobile"
## [1] "imperforate" "perforate"
## [1] "impermeable" "permeable"
## [1] "inflated"
                   "uninflated"
## [1] "insensitive toward" "sensitive toward"
## [1] "insoluble in" "soluble in"
## [1] "insufficient" "sufficient"
## [1] "insusceptible toward" "susceptible toward"
## [1] "invariant" "variant"
## [1] "invariant color" "variant color"
## [1] "invariant shape" "variant shape"
## [1] "invariant temperature" "variant temperature"
## [1] "irregular duration" "regular duration"
## [1] "irregular sleep pattern" "regular sleep pattern"
## [1] "irregular spatial pattern" "regular spatial pattern"
## [1] "lumenized" "unlumenized"
## [1] "magnetic"
                     "non-magnetic"
## [1] "male fertile" "male sterile"
## [1] "male semi-fertile" "male semi-sterile"
## [1] "necessary (continuant)" "unnecessary (continuant)"
## [1] "necessary (occurrent)" "unnecessary (occurrent)"
## [1] "negative charge" "positive charge"
## [1] "non-porous" "porous"
## [1] "non-potable" "potable"
## [1] "non-progressive" "progressive"
## [1] "old" "young"
## [1] "opaque" "transparent"
```

```
## [1] "ornamented"
                      "unornamented"
## [1] "pigmented"
                     "unpigmented"
                         "unresponsive to"
## [1] "responsive to"
## [1] "rough" "smooth"
## [1] "serrated"
                    "unserrated"
## [1] "stratified"
                      "unstratified"
## [1] "structured"
                      "unstructured"
## [1] ""
## [1] "IN MINE ONLY:"
## [1] "abaxial to" "axial to"
## [1] "acyclic cyclicity" "cyclic cyclicity"
## [1] "alobate" "lobate"
## [1] "anaplastic" "aplastic"
## [1] "aneuploid" "euploid"
## [1] "asymmetrical" "symmetrical"
## [1] "atonicity" "tonicity"
## [1] "complex" "simple"
## [1] "curved dorsal" "curved ventral"
## [1] "dark blue" "light blue"
## [1] "dark brown" "light brown"
## [1] "dark cyan" "light cyan"
## [1] "dark green" "light green"
## [1] "dark grey" "light grey"
## [1] "dark magenta" "light magenta"
## [1] "dark orange" "light orange"
## [1] "dark phase" "light phase"
## [1] "dark purple" "light purple"
## [1] "dark red" "light red"
## [1] "dark red brown" "light red brown"
## [1] "dark violet" "light violet"
## [1] "dark yellow" "light yellow"
## [1] "dark yellow brown" "light yellow brown"
## [1] "decreased linear velocity" "increased linear velocity"
## [1] "decreased propagation velocity" "increased propagation velocity"
## [1] "decreased proportionality to" "increased proportionality to"
## [1] "decreased spatial extent of a process"
## [2] "increased spatial extent of a process"
## [1] "desaturated blue" "saturated blue"
## [1] "desaturated brown" "saturated brown"
## [1] "desaturated cyan" "saturated cyan"
## [1] "desaturated green" "saturated green"
## [1] "desaturated magenta" "saturated magenta"
## [1] "desaturated orange" "saturated orange"
## [1] "desaturated purple" "saturated purple"
## [1] "desaturated red" "saturated red"
## [1] "desaturated violet" "saturated violet"
## [1] "desaturated yellow" "saturated yellow"
## [1] "disposition" "position"
## [1] "dorsal orientation" "ventral orientation"
## [1] "dorsal to" "ventral to"
```

```
## [1] "dystonicity" "tonicity"
## [1] "glutinous"
                       "non-glutinous"
## [1] "ictal"
                  "non-ictal"
## [1] "immobile relative to" "mobile relative to"
## [1] "nonpolar polarity" "polar polarity"
## [1] "obsolete backcross fertile" "obsolete backcross sterile"
## [1] "obsolete decreased action potential" "obsolete increased action potential"
## [1] "obsolete decreased consumption" "obsolete increased consumption"
## [1] "obsolete decreased percentage" "obsolete increased percentage"
## [1] "obsolete decreased relaxation" "obsolete increased relaxation"
## [1] "obsolete f1 fertile" "obsolete f1 sterile"
## [1] "obsolete f2 fertile" "obsolete f2 sterile"
## [1] "obsolete high" "obsolete low"
## [1] "obsolete high enzyme function value" "obsolete low enzyme function value"
## [1] "obsolete high percentage" "obsolete low percentage"
## [1] "obsolete high yield" "obsolete low yield"
## [1] "obsolete intercross fertile" "obsolete intercross sterile"
## [1] "obsolete negative regulation" "obsolete positive regulation"
## [1] "splayed dorsal" "splayed ventral"
## [1] ""
## [1] "IN THE CURRENT PAIRS ONLY:"
## [1] "anterodistal orientation" "anterodorsal orientation"
## [1] "auxotrophic" "prototrophic"
## [1] "behavioural active"
                            "behavioural inactive"
## [1] "biconcave" "biconvex"
## [1] "concave" "convex"
## [1] "concave 3-D shape" "convex 3-D shape"
## [1] "distal orientation" "dorsal orientation"
## [1] "distal to" "dorsal to"
## [1] "east" "west"
## [1] "fused with"
                      "unfused from"
## [1] "has extra parts of type" "has fewer parts of type"
## [1] "hydrophilic" "hydrophobic"
## [1] "hydrophilicity" "hydrophobicity"
## [1] "hyperelliptic" "hypoelliptic"
## [1] "hyperplastic" "hypoplastic"
## [1] "hyperresponsive to" "hyporesponsive to"
## [1] "hypertrophic" "hypotrophic"
## [1] "hypertrophic growth" "hypotrophic growth"
## [1] "increased virulence" "reduced virulence"
## [1] "large (value)" "small (value)"
## [1] "north" "south"
## [1] "paedomorphic growth" "peramorphic growth"
## [1] "photoinsensitive" "photosensitive"
## [1] "posterodistal orientation" "posterodorsal orientation"
## [1] "recurrent" "sporadic"
## [1] "relative large (value)" "relative small (value)"
## [1] "resistance to"
                             "susceptibility toward"
```

Comparison to Chris et al.'s stuff



```
## [1] "Mine:"
## # A tibble: 302 x 2
##
      term01
                term02
##
      <chr>>
                 <chr>>
  1 HP:0030469 HP:0008275
##
## 2 HP:0100737 HP:0100736
  3 HP:0025090 HP:0025129
## 4 HP:0031199 HP:0031197
## 5 HP:0011790 HP:0011791
## 6 HP:0012388 HP:0004469
## 7 HP:0100282 HP:0100281
## 8 HP:0012451 HP:0012450
## 9 HP:0032675 HP:0032676
## 10 HP:0011880 HP:0005520
## # ... with 292 more rows
## [1] "Chris's:"
## # A tibble: 184 x 2
##
      term01
                term02
##
      <chr>
                 <chr>
## 1 HP:0000098 HP:0004322
```

```
## 2 HP:0000103 HP:0011037
## 3 HP:0000154 HP:0000160
## 4 HP:0000158 HP:0000171
## 5 HP:0000188 HP:0011341
## 6 HP:0000239 HP:0005486
## 7 HP:0000253 HP:0004481
## 8 HP:0000274 HP:0100729
## 9 HP:0000275 HP:0000283
## 10 HP:0000276 HP:0011219
## # ... with 174 more rows
OK, we know the intersection/complements, so let's see what Chris's opposites look like, exactly.
## Warning: package 'ontologyIndex' was built under R version 4.0.2
## [1] "FOUND BY BOTH ME AND CHRIS:"
## [1] "Decreased activity of mitochondrial respiratory chain
                                                           Increased activity of mitochondrial re
## [1] "Decreased circulating ACTH level" Increased circulating ACTH level"
## [1] "Decreased circulating antibody level" Increased circulating antibody level"
## [1] "Decreased HDL cholesterol concentration
                                               Increased HDL cholesterol concentration"
## [1] "Decreased inflammatory response" Increased inflammatory response"
## [1] "Decreased mitochondrial number
                                       Increased mitochondrial number"
## [1] "Decreased pineal volume
                             Increased pineal volume"
## [1] "Decreased size of nasopharyngeal adenoids
                                                Increased size of nasopharyngeal adenoids"
## [1] "Decreased urinary sulfate
                                  Increased urinary sulfate"
## [1] "Decreased urinary urate
                                Increased urinary urate"
## [1] "Large basal ganglia Small basal ganglia"
## [1] "Large humeral epiphyses Small humeral epiphyses"
## [1] "Large radial epiphyses
                               Small radial epiphyses"
## [1] "Large sella turcica
                            Small sella turcica"
## [1] "FOUND BY ME, NOT BY CHRIS:"
## [1] ""
## [1] "Abnormal dark-adapted electroretinogram Abnormal light-adapted electroretinogram"
## [1] "Abnormal large intestinal mucosa morphology
                                                  Abnormal small intestinal mucosa morphology"
## [1] "Activating thyroid-stimulating hormone receptor defect
                                                            Inactivating thyroid-stimulating hor
## [1] "Acute bronchitis
                         Chronic bronchitis"
## [1] "Acute colitis
                       Chronic colitis"
## [1] "Acute constipation
                            Chronic constipation"
## [1] "Acute cutaneous wound
                              Chronic cutaneous wound"
## [1] "Acute disseminated intravascular coagulation
                                                   Chronic disseminated intravascular coagulation
## [1] "Acute hepatic failure
                              Chronic hepatic failure"
## [1] "Acute hepatitis Chronic hepatitis"
## [1] "Acute leukemia
                       Chronic leukemia"
## [1] "Acute myelomonocytic leukemia
                                   Chronic myelomonocytic leukemia"
```

```
## [1] "Acute otitis media
                               Chronic otitis media"
## [1] "Acute pancreatitis
                               Chronic pancreatitis"
## [1] "Acute respiratory acidosis
                                       Chronic respiratory acidosis"
## [1] "Acute sinusitis
                            Chronic sinusitis"
## [1] "Acute tubulointerstitial nephritis
                                               Chronic tubulointerstitial nephritis"
## [1] "Acute
                  Chronic"
## [1] "Adenocarcinoma of the large intestine
                                                  Adenocarcinoma of the small intestine"
## [1] "Affected
                     Unaffected"
## [1] "Agranulocytosis
                            Granulocytosis"
## [1] "Alobar holoprosencephaly
                                     Lobar holoprosencephaly"
## [1] "Anhidrotic ectodermal dysplasia
                                            Hidrotic ectodermal dysplasia"
## [1] "Asymmetric peripheral demyelination
                                                Symmetric peripheral demyelination"
                           Tonic seizure"
## [1] "Atonic seizure
                                     Typical absence seizure"
## [1] "Atypical absence seizure
## [1] "Atypical absence status epilepticus
                                                Typical absence status epilepticus"
## [1] "Cleft hard palate
                              Cleft soft palate"
                          Open comedo"
## [1] "Closed comedo
## [1] "Communicating hydrocephalus
                                        Noncommunicating hydrocephalus"
## [1] "Complete atrioventricular canal defect
                                                   Partial atrioventricular canal defect"
## [1] "Complete breech presentation
                                         Incomplete breech presentation"
## [1] "Complete congenital stationary night blindness
                                                           Incomplete congenital stationary night blind
## [1] "Complete diaphragmatic absence of pericardium
                                                          Partial diaphragmatic absence of pericardium"
## [1] "Complete duplication of the 1st metatarsal
                                                       Partial duplication of the 1st metatarsal"
## [1] "Complete duplication of the distal phalanges of the hand
                                                                      Partial duplication of the distal
## [1] "Complete duplication of the distal phalanx of the 2nd finger
                                                                          Partial duplication of the dis
## [1] "Complete duplication of the distal phalanx of the 2nd toe
                                                                       Partial duplication of the distal
## [1] "Complete duplication of the distal phalanx of the 3rd finger
                                                                          Partial duplication of the dis
## [1] "Complete duplication of the distal phalanx of the 3rd toe
                                                                       Partial duplication of the distal
## [1] "Complete duplication of the distal phalanx of the 4th finger
                                                                          Partial duplication of the dis
## [1] "Complete duplication of the distal phalanx of the 4th toe
                                                                       Partial duplication of the distal
## [1] "Complete duplication of the distal phalanx of the 5th finger
                                                                          Partial duplication of the dis
## [1] "Complete duplication of the distal phalanx of the 5th toe
                                                                       Partial duplication of the distal
## [1] "Complete duplication of the distal phalanx of the hallux
                                                                      Partial duplication of the distal
## [1] "Complete duplication of the middle phalanges of the hand
                                                                      Partial duplication of the middle
## [1] "Complete duplication of the middle phalanx of the 2nd finger
                                                                          Partial duplication of the mid-
## [1] "Complete duplication of the middle phalanx of the 2nd toe
                                                                       Partial duplication of the middle
## [1] "Complete duplication of the middle phalanx of the 3rd finger
                                                                          Partial duplication of the mid-
## [1] "Complete duplication of the middle phalanx of the 3rd toe
                                                                       Partial duplication of the middle
## [1] "Complete duplication of the middle phalanx of the 4th finger
                                                                          Partial duplication of the mid-
## [1] "Complete duplication of the middle phalanx of the 4th toe
                                                                       Partial duplication of the middle
## [1] "Complete duplication of the middle phalanx of the 5th finger
                                                                          Partial duplication of the mid-
## [1] "Complete duplication of the middle phalanx of the 5th toe
                                                                       Partial duplication of the middle
## [1] "Complete duplication of the phalanges of the 2nd finger
                                                                     Partial duplication of the phalange
## [1] "Complete duplication of the phalanges of the 3rd finger
                                                                     Partial duplication of the phalange
## [1] "Complete duplication of the phalanges of the 4th finger
                                                                     Partial duplication of the phalange
## [1] "Complete duplication of the phalanges of the 5th finger
                                                                     Partial duplication of the phalange
## [1] "Complete duplication of the proximal phalanges of the hand
                                                                        Partial duplication of the proximal
## [1] "Complete duplication of the proximal phalanx of the 2nd finger
                                                                            Partial duplication of the p
## [1] "Complete duplication of the proximal phalanx of the 2nd toe
                                                                         Partial duplication of the prox
## [1] "Complete duplication of the proximal phalanx of the 3rd finger
                                                                            Partial duplication of the p
## [1] "Complete duplication of the proximal phalanx of the 3rd toe
                                                                         Partial duplication of the prox
## [1] "Complete duplication of the proximal phalanx of the 4th finger
                                                                            Partial duplication of the partial
## [1] "Complete duplication of the proximal phalanx of the 4th toe
                                                                         Partial duplication of the prox
## [1] "Complete duplication of the proximal phalanx of the 5th finger
                                                                            Partial duplication of the p
```

```
## [1] "Complete duplication of the proximal phalanx of the 5th toe
                                                                      Partial duplication of the prox
## [1] "Complete duplication of the proximal phalanx of the hallux
                                                                      Partial duplication of the proximation
## [1] "Complete duplication of thumb phalanx
                                                 Partial duplication of thumb phalanx"
## [1] "Complete left sided absence of pericardium
                                                      Partial left sided absence of pericardium"
## [1] "Complete right sided absence of pericardium
                                                       Partial right sided absence of pericardium"
## [1] "Complex febrile seizure
                                   Simple febrile seizure"
## [1] "Complex renal cyst
                              Simple renal cyst"
## [1] "Conjugated hyperbilirubinemia
                                         Unconjugated hyperbilirubinemia"
## [1] "Cyclophoria" Incyclophoria"
## [1] "Cyclotropia
                       Incyclotropia"
## [1] "Decreased adiponectin level
                                       Increased adiponectin level"
## [1] "Decreased adipose tissue
                                     Increased adipose tissue"
## [1] "Decreased antimullerian hormone level
                                                  Increased antimullerian hormone level"
## [1] "Decreased Arden ratio of electrooculogram
                                                      Increased Arden ratio of electrooculogram"
## [1] "Decreased basophil count
                                     Increased basophil count"
## [1] "Decreased biotinidase level
                                        Increased biotinidase level"
## [1] "Decreased blood drug concentration
                                               Increased blood drug concentration"
## [1] "Decreased body fat percentage
                                         Increased body fat percentage"
## [1] "Decreased body mass index
                                     Increased body mass index"
## [1] "Decreased body weight
                                  Increased body weight"
## [1] "Decreased circulating 18-hydroxycortisone level
                                                            Increased circulating 18-hydroxycortisone 1
## [1] "Decreased circulating A-type natriuretic peptide level
                                                                   Increased circulating A-type natriur
## [1] "Decreased circulating androgen level
                                             Increased circulating androgen level"
## [1] "Decreased circulating beta-2-microglobulin level
                                                             Increased circulating beta-2-microglobulin
## [1] "Decreased circulating beta-C-terminal telopeptide level
                                                                    Increased circulating beta-C-termin
## [1] "Decreased circulating ceruloplasmin concentration
                                                              Increased circulating ceruloplasmin conce
## [1] "Decreased circulating chylomicron concentration
                                                            Increased circulating chylomicron concentra
## [1] "Decreased circulating copper concentration
                                                      Increased circulating copper concentration"
## [1] "Decreased circulating corticosterone level
                                                       Increased circulating corticosterone level"
## [1] "Decreased circulating cortisol level
                                                 Increased circulating cortisol level"
## [1] "Decreased circulating free fatty acid level
                                                        Increased circulating free fatty acid level"
## [1] "Decreased circulating free T3
                                          Increased circulating free T3"
## [1] "Decreased circulating free T4 level
                                                Increased circulating free T4 level"
## [1] "Decreased circulating globulin level
                                                 Increased circulating globulin level"
## [1] "Decreased circulating gonadotropin level
                                                     Increased circulating gonadotropin level"
## [1] "Decreased circulating heparan sulfate level
                                                       Increased circulating heparan sulfate level"
## [1] "Decreased circulating IgA level Increased circulating IgA level"
## [1] "Decreased circulating IgG level
                                            Increased circulating IgG level"
## [1] "Decreased circulating IgG1 level
                                            Increased circulating IgG1 level"
## [1] "Decreased circulating IgG2 level
                                            Increased circulating IgG2 level"
## [1] "Decreased circulating IgG3 level
                                             Increased circulating IgG3 level"
## [1] "Decreased circulating IgG4 level
                                             Increased circulating IgG4 level"
## [1] "Decreased circulating osteocalcin level
                                                    Increased circulating osteocalcin level"
## [1] "Decreased circulating progesterone
                                               Increased circulating progesterone"
## [1] "Decreased circulating purine concentration
                                                       Increased circulating purine concentration"
## [1] "Decreased circulating renin level
                                              Increased circulating renin level"
## [1] "Decreased circulating selenium concentration
                                                         Increased circulating selenium concentration"
## [1] "Decreased circulating T4 level
                                           Increased circulating T4 level"
## [1] "Decreased complex N-glycan level
                                            Increased complex N-glycan level"
## [1] "Decreased core 1 O-glycan level
                                            Increased core 1 O-glycan level"
## [1] "Decreased corneal thickness
                                        Increased corneal thickness"
## [1] "Decreased CSF alanine concentration
                                                Increased CSF alanine concentration"
                                                Increased CSF albumin concentration"
## [1] "Decreased CSF albumin concentration
## [1] "Decreased CSF arginine concentration
                                                Increased CSF arginine concentration"
```

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## [1] "Decreased CSF glutamate concentration
                                               Increased CSF glutamate concentration"
## [1] "Decreased CSF glutamine concentration
                                               Increased CSF glutamine concentration"
                                               Increased CSF histidine concentration"
## [1] "Decreased CSF histidine concentration
## [1] "Decreased CSF isoleucine concentration
                                                Increased CSF isoleucine concentration"
## [1] "Decreased CSF leucine concentration
                                             Increased CSF leucine concentration"
## [1] "Decreased CSF lysine concentration
                                             Increased CSF lysine concentration"
## [1] "Decreased CSF phenylalanine concentration
                                                   Increased CSF phenylalanine concentration"
## [1] "Decreased CSF protein
                                Increased CSF protein"
                                             Increased CSF serine concentration"
## [1] "Decreased CSF serine concentration
## [1] "Decreased CSF threonine concentration
                                               Increased CSF threonine concentration"
## [1] "Decreased CSF tyrosine concentration
                                              Increased CSF tyrosine concentration"
## [1] "Decreased CSF valine concentration
                                            Increased CSF valine concentration"
## [1] "Decreased CSF/serum albumin ratio
                                            Increased CSF/serum albumin ratio"
## [1] "Decreased cystatin C level
                                     Increased cystatin C level"
## [1] "Decreased DLCO
                         Increased DLCO"
## [1] "Decreased female libido
                                  Increased female libido"
## [1] "Decreased femoral torsion
                                    Increased femoral torsion"
## [1] "Decreased fetal movement
                                   Increased fetal movement"
## [1] "Decreased fibular diameter
                                     Increased fibular diameter"
## [1] "Decreased fucosylation of N-linked protein glycosylation
                                                                 Increased fucosylation of N-linked
## [1] "Decreased fucosylation of O-linked protein glycosylation
                                                                  Increased fucosylation of O-linked
## [1] "Decreased glucagon level
                                   Increased glucagon level"
## [1] "Decreased glucose-6-phosphate dehydrogenase level in blood
                                                                   Increased glucose-6-phosphate de
## [1] "Decreased glucose-6-phosphate dehydrogenase level in dried blood spot Increased glucose-6-p
## [1] "Decreased glucose-6-phosphate dehydrogenase level in red blood cells
                                                                          Increased glucose-6-ph
## [1] "Decreased glucose-6-phosphate dehydrogenase level in tissue
                                                                    Increased glucose-6-phosphate d
## [1] "Decreased HDL2a concentration
                                        Increased HDL2a concentration"
## [1] "Decreased HDL2b concentration
                                        Increased HDL2b concentration"
## [1] "Decreased HDL3a concentration
                                        Increased HDL3a concentration"
## [1] "Decreased HDL3b concentration
                                        Increased HDL3b concentration"
## [1] "Decreased HDL3c concentration
                                        Increased HDL3c concentration"
## [1] "Decreased head circumference
                                       Increased head circumference"
## [1] "Decreased heart rate variability
                                          Increased heart rate variability"
## [1] "Decreased helper T cell proportion
                                            Increased helper T cell proportion"
## [1] "Decreased hemoglobin concentration
                                            Increased hemoglobin concentration"
## [1] "Decreased hepatic echogenicity
                                         Increased hepatic echogenicity"
## [1] "Decreased high-mannose N-glycan level
                                               Increased high-mannose N-glycan level"
## [1] "Decreased immunoglobulin level in body fluid
                                                      Increased immunoglobulin level in body fluid"
## [1] "Decreased incisura length
                                    Increased incisura length"
## [1] "Decreased intestinal transit time
                                           Increased intestinal transit time"
## [1] "Decreased intracranial pressure
                                          Increased intracranial pressure"
## [1] "Decreased LDL cholesterol concentration
                                                 Increased LDL cholesterol concentration"
## [1] "Decreased level of carnosine in blood
                                               Increased level of carnosine in blood"
## [1] "Decreased level of GABA in serum
                                           Increased level of GABA in serum"
                                                       Increased level of platelet-activating factor
## [1] "Decreased level of platelet-activating factor
## [1] "Decreased libido
                           Increased libido"
## [1] "Decreased male libido
                                Increased male libido"
## [1] "Decreased mannose-binding protein level
                                                 Increased mannose-binding protein level"
## [1] "Decreased mannosylation of N-linked protein glycosylation Increased mannosylation of N-link
## [1] "Decreased mean corpuscular hemoglobin concentration Increased mean corpuscular hemoglobin c
```

[1] "Decreased monosialylated core 1 0-glycan level Increased monosialylated core 1 0-glycan lev

[1] "Decreased mean corpuscular volume Increased mean corpuscular volume"

```
## [1] "Decreased muscle glycogen content" Increased muscle glycogen content"
## [1] "Decreased phosphoribosylpyrophosphate synthetase level Increased phosphoribosylpyrophosphat
                                                             Increased pituitary glycoprote
## [1] "Decreased pituitary glycoprotein hormone alpha subunit level
## [1] "Decreased proportion of CD4+CD25+ regulatory T cells Increased proportion of CD4+CD25+ regu
## [1] "Decreased proportion of CD4-positive T cells  
Increased proportion of CD4-positive T cells"
## [1] "Decreased proportion of central memory CD4-positive, alpha-beta T cells Increased proportion
## [1] "Decreased proportion of central memory CD8-positive, alpha-beta T cells
                                                                     Increased proportion
## [1] "Decreased proportion of class-switched memory B cells Increased proportion of class-switche
## [1] "Decreased proportion of effector memory CD8-positive, alpha-beta T cells Increased proporti
## [1] "Decreased proportion of gamma-delta T cells Increased proportion of gamma-delta T cells"
## [1] "Decreased proportion of immature B cells
                                         Increased proportion of immature B cells"
## [1] "Decreased proportion of immature gamma-delta T cells
                                                     Increased proportion of immature gamma
## [1] "Decreased proportion of marginal zone B cells
                                                Increased proportion of marginal zone B cells
Increased proportion of memory T cells"
## [1] "Decreased proportion of memory T cells
## [1] "Decreased proportion of naive B cells
                                         Increased proportion of naive B cells"
## [1] "Decreased proportion of naive CD4 T cells
                                             Increased proportion of naive CD4 T cells"
## [1] "Decreased proportion of naive CD8 T cells
                                             Increased proportion of naive CD8 T cells"
## [1] "Decreased proportion of naive T cells
                                        Increased proportion of naive T cells"
## [1] "Decreased proportion of plasmablasts
                                        Increased proportion of plasmablasts"
## [1] "Decreased proportion of transitional B cells
                                               Increased proportion of transitional B cells"
## [1] "Decreased proportion of unswitched memory B cells
                                                   Increased proportion of unswitched memory
## [1] "Decreased QRS voltage
                            Increased QRS voltage"
## [1] "Decreased red blood cell count
                                 Increased red blood cell count"
## [1] "Decreased resting energy expenditure" Increased resting energy expenditure"
## [1] "Decreased serum estradiol
                               Increased serum estradiol"
## [1] "Decreased serum estriol
                              Increased serum estriol"
## [1] "Decreased serum estrone
                              Increased serum estrone"
## [1] "Decreased serum ferritin
                              Increased serum ferritin"
## [1] "Decreased serum insulin-like growth factor 1
                                               Increased serum insulin-like growth factor 1"
## [1] "Decreased serum iron
                           Increased serum iron"
## [1] "Decreased serum leptin
                             Increased serum leptin"
## [1] "Decreased serum testosterone level
                                       Increased serum testosterone level"
## [1] "Decreased serum zinc
                           Increased serum zinc"
## [1] "Decreased sialylated N-glycan level
                                     Increased sialylated N-glycan level"
## [1] "Decreased sialylation of N-linked protein glycosylation
                                                        Increased sialylation of N-linked p
                                    Increased size of the clitoris"
## [1] "Decreased size of the clitoris
## [1] "Decreased skull ossification
                                  Increased skull ossification"
## [1] "Decreased superoxide dismutase level
                                        Increased superoxide dismutase level"
                           Increased T3/T4 ratio"
## [1] "Decreased T3/T4 ratio
## [1] "Decreased total hemolytic complement activity
                                                Increased total hemolytic complement activity
## [1] "Decreased uridine diphosphate glucose-4-epimerase level in red blood cells
                                                                         Increased uridin
## [1] "Decreased urinary 1-methylhistidine"
## [1] "Decreased urinary 3-methylhistidine"
Increased urinary 3-methylhistidine"
                                         Increased urinary copper concentration"
## [1] "Decreased urinary copper concentration
## [1] "Decreased urine alpha-ketoglutarate concentration Increased urine alpha-ketoglutarate conce
```

```
## [1] "Decreased urine urobilinogen
                                        Increased urine urobilinogen"
## [1] "Decreased VLDL cholesterol concentration
                                                    Increased VLDL cholesterol concentration"
## [1] "Decreased waist to hip ratio
                                        Increased waist to hip ratio"
## [1] "Disproportionate short stature
                                         Proportionate short stature"
## [1] "Disproportionate tall stature
                                         Proportionate tall stature"
## [1] "Dysgerminoma
                        Germinoma"
## [1] "Early chronotype
                            Late chronotype"
## [1] "Early inspiratory crackles
                                      Late inspiratory crackles"
## [1] "Early spermatogenesis maturation arrest
                                                   Late spermatogenesis maturation arrest"
## [1] "Fast-growing nails
                              Slow-growing nails"
## [1] "Happy demeanor
                         Unhappy demeanor"
## [1] "Heavy-chain paraproteinemia
                                       Light-chain paraproteinemia"
## [1] "High anterior hairline
                                  Low anterior hairline"
## [1] "High insertion of columella
                                       Low insertion of columella"
## [1] "High maternal serum alpha-fetoprotein
                                                Low maternal serum alpha-fetoprotein"
## [1] "High maternal serum chorionic gonadotropin" Low maternal serum chorionic gonadotropin"
## [1] "High posterior hairline
                                  Low posterior hairline"
## [1] "High pulse pressure
                             Low pulse pressure"
## [1] "High serum calcifediol
                                 Low serum calcifediol"
## [1] "High serum calcitriol
                                 Low serum calcitriol"
## [1] "High-frequency sensorineural hearing impairment
                                                           Low-frequency sensorineural hearing impairm
## [1] "High-output congestive heart failure" Low-output congestive heart failure"
## [1] "Hodgkin lymphoma" Non-Hodgkin lymphoma"
## [1] "Ketotic hypoglycemia
                                Nonketotic hypoglycemia"
## [1] "Large cell lung carcinoma
                                     Small cell lung carcinoma"
## [1] "Large earlobe
                         Small earlobe"
## [1] "Large face
                      Small face"
## [1] "Large for gestational age
                                     Small for gestational age"
## [1] "Large foramen magnum
                                Small foramen magnum"
## [1] "Large forehead
                          Small forehead"
## [1] "Large intestinal polyposis
                                      Small intestinal polyposis"
## [1] "Large placenta
                          Small placenta"
## [1] "Large vessel vasculitis
                                  Small vessel vasculitis"
## [1] "Medial calcification of large arteries Medial calcification of small arteries"
## [1] "Medullary thyroid carcinoma
                                       Non-medullary thyroid carcinoma"
## [1] "Monoclonal elevation of IgG heavy chain
                                                Monoclonal elevation of IgG light chain"
## [1] "Neoplasm of the large intestine
                                           Neoplasm of the small intestine"
## [1] "Non-obstructive azoospermia
                                       Obstructive azoospermia"
## [1] "Non-restrictive ventricular septal defect
                                                     Restrictive ventricular septal defect"
## [1] "Non-secretory adrenocortical adenoma
                                                Secretory adrenocortical adenoma"
## [1] "Non-small cell lung carcinoma
                                         Small cell lung carcinoma"
## [1] "Nonopposable triphalangeal thumb
                                            Opposable triphalangeal thumb"
## [1] "Nonproductive cough Productive cough"
## [1] "Nonprogressive cerebellar ataxia
                                            Progressive cerebellar ataxia"
## [1] "Nonprogressive encephalopathy
                                         Progressive encephalopathy"
                                      Progressive visual loss"
## [1] "Nonprogressive visual loss
## [1] "Nonprogressive
                          Progressive"
## [1] "obsolete Decreased proportion of CD4+ central memory cells
                                                                      obsolete Increased proportion of
```

14

Secondary Caesarian section"

Secondary hypercortisolism"

Secondary hyperaldosteronism"

Tertiary hyperparathyroidism"

Secondary amenorrhea"

[1] "Primary hyperparathyroidism Secondary hyperparathyroidism"

[1] "Primary amenorrhea

[1] "Primary Caesarian section

[1] "Primary hypercortisolism

[1] "Primary hyperaldosteronism

[1] "Secondary hyperparathyroidism

```
## [1] "Submucous cleft hard palate
                                        Submucous cleft soft palate"
## [1] "Undetectable dark-adapted electroretinogram
                                                        Undetectable light-adapted electroretinogram"
## [1] "FOUND BY CHRIS, NOT BY ME:"
## [1] ""
## [1] "Tall stature
                         Short stature"
## [1] "Polyuria
                     Decreased urine output"
## [1] "Wide mouth
                       Narrow mouth"
## [1] "Macroglossia
                         Microglossia"
## [1] "Short upper lip
                            Long upper lip"
## [1] "Large fontanelles
                              Small fontanelle"
## [1] "Progressive microcephaly
                                     Progressive macrocephaly"
## [1] "Small face
                       Large face"
## [1] "Narrow face
                        Broad face"
## [1] "Long face
                      Short face"
## [1] "Hypertelorism
                          Hypotelorism"
## [1] "Short philtrum
                           Long philtrum"
## [1] "Hypoplasia of the maxilla
                                      Hyperplasia of the maxilla"
## [1] "Short chin
                       Tall chin"
## [1] "Broad forehead
                           Narrow forehead"
## [1] "Small forehead
                           Large forehead"
## [1] "Wide nose
                      Narrow nose"
## [1] "Short neck
                       Long neck"
## [1] "Microcornea
                        Megalocornea"
## [1] "Long eyelashes
                           Short eyelashes"
## [1] "Thick eyebrow
                          Thin eyebrow"
## [1] "Long palpebral fissure
                                   Short palpebral fissure"
## [1] "Hypodontia
                       Increased number of teeth"
## [1] "Microdontia
                        Macrodontia"
## [1] "Hypoplasia of the thymus
                                     Thymus hyperplasia"
## [1] "Hypothyroidism
                           Hyperthyroidism"
## [1] "Hypertension
                         Hypotension"
## [1] "Hypoparathyroidism
                               Hyperparathyroidism"
## [1] "Increased circulating gonadotropin level
                                                     Decreased circulating gonadotropin level"
## [1] "Hyperinsulinemia
                             Hypoinsulinemia"
## [1] "Increased circulating renin level
                                              Decreased circulating renin level"
## [1] "Hyperaldosteronism
                               Decreased circulating aldosterone level"
## [1] "Parathyroid hypoplasia
                                   Parathyroid hyperplasia"
## [1] "Long clavicles
                           Short clavicles"
## [1] "Hypertrichosis
                           obsolete Hypotrichosis"
## [1] "Broad palm
                       Narrow palm"
## [1] "Large hands
                        Small hand"
## [1] "Short umbilical cord
                                 Long umbilical cord"
## [1] "Hyporeflexia
                         Hyperreflexia"
## [1] "Limitation of joint mobility
                                         Joint hypermobility"
## [1] "Onychogryposis
                           Thin nail"
## [1] "Broad nail
                       Narrow nail"
## [1] "Short toe
                      Long toe"
                        Short hallux"
## [1] "Long hallux
## [1] "Leukopenia
                       Leukocytosis"
```

Hyperglycemia"

[1] "Hypoglycemia

```
## [1] "Hypophosphatemia
                             Hyperphosphatemia"
                          Hypouricemia"
## [1] "Hyperuricemia
## [1] "Hypercalciuria
                           Hypocalciuria"
## [1] "Hyperkalemia
                         Hypokalemia"
## [1] "Hyperglycinemia
                            Hypoglycinemia"
## [1] "Hypertriglyceridemia
                                 Hypotriglyceridemia"
## [1] "Increased CSF lactate
                                  Reduced CSF lactate"
## [1] "Pancreatic hypoplasia
                                  Pancreatic hyperplasia"
## [1] "Small foramen magnum
                                 Large foramen magnum"
## [1] "Thickened calvaria
                               Thin calvarium"
## [1] "Lymphadenopathy
                            Lymph node hypoplasia"
## [1] "Hypoventilation
                            Hyperventilation"
## [1] "Decreased circulating total IgM
                                            Increased circulating IgM level"
## [1] "Hypocalcemia
                         Hypercalcemia"
## [1] "Hyponatremia
                         Hypernatremia"
## [1] "Hyperchloriduria
                             Hypochloriduria"
## [1] "Hypomagnesemia
                           Hypermagnesemia"
## [1] "Hypoalbuminemia
                            Hyperalbuminemia"
## [1] "Hyperlipidemia
                           Hypolipidemia"
## [1] "Increased urinary potassium
                                        Decreased urinary potassium"
## [1] "Hyperphosphaturia
                              Hypophosphaturia"
## [1] "Hypochloremia
                          Hyperchloremia"
## [1] "Increased circulating cortisol level
                                                 Decreased circulating cortisol level"
## [1] "Hypercholesterolemia
                                 Hypocholesterolemia"
## [1] "Elevated alkaline phosphatase
                                          Low alkaline phosphatase"
## [1] "Hypermethioninemia
                               Hypomethioninemia"
                                            Decreased circulating IgG level"
## [1] "Increased circulating IgG level
## [1] "Elevated serum creatinine
                                      Decreased serum creatinine"
## [1] "Increased serum ferritin
                                     Decreased serum ferritin"
## [1] "CNS hypomyelination
                                CNS hypermyelination"
## [1] "Short palm
                       Long palm"
## [1] "Increased body weight
                                  Decreased body weight"
## [1] "Increased skull ossification
                                         Decreased skull ossification"
## [1] "Reduced bone mineral density
                                         Increased bone mineral density"
## [1] "Increased circulating purine concentration
                                                       Decreased circulating purine concentration"
## [1] "Biconcave vertebral bodies
                                       Biconvex vertebral bodies"
## [1] "Short middle phalanx of finger
                                           Long middle phalanx of finger"
## [1] "Thyroid hypoplasia
                               Thyroid hyperplasia"
## [1] "Long proximal phalanx of finger
                                            Short proximal phalanx of finger"
## [1] "Small placenta
                           Large placenta"
## [1] "Broad femoral neck
                               Narrow femoral neck"
                                       Peripheral hypermyelination"
## [1] "Peripheral hypomyelination
                                          Generalized hypopigmentation"
## [1] "Generalized hyperpigmentation
## [1] "Long upper eyelashes
                                 Short upper eyelashes"
## [1] "Macular hypopigmentation
                                     Macular hyperpigmentation"
## [1] "Foveal hyperpigmentation
                                     Foveal hypopigmentation"
## [1] "Increased muscle glycogen content
                                              Decreased muscle glycogen content"
## [1] "Increased adipose tissue
                                     Decreased adipose tissue"
## [1] "Enlarged epiphysis of the distal phalanx of the 5th finger
                                                                        Small epiphysis of the distal ph
## [1] "Enlarged epiphysis of the proximal phalanx of the 5th finger
                                                                          Small epiphysis of the proxima
## [1] "Enlarged epiphysis of the middle phalanx of the 5th finger
                                                                        Small epiphysis of the middle ph
## [1] "Enlarged epiphysis of the middle phalanx of the 4th finger
                                                                        Small epiphysis of the middle ph
## [1] "Enlarged epiphysis of the distal phalanx of the 4th finger
                                                                        Small epiphysis of the distal ph
```

[1] "Hyperammonemia

Hypoammonemia"

```
## [1] "Enlarged epiphysis of the proximal phalanx of the 4th finger
                                                          Small epiphysis of the proxima
## [1] "Enlarged epiphysis of the middle phalanx of the 3rd finger Small epiphysis of the middle ph
## [1] "Enlarged epiphysis of the distal phalanx of the 3rd finger
                                                          Small epiphysis of the distal ph
## [1] "Enlarged epiphyses of the 5th finger Small epiphyses of the 5th finger"
## [1] "Enlarged epiphyses of the 4th finger Small epiphyses of the 4th finger"
## [1] "Enlarged epiphyses of the 3rd finger Small epiphyses of the 3rd finger"
## [1] "Enlarged epiphyses of the 2nd finger Small epiphyses of the 2nd finger"
## [1] "Enlarged epiphysis of the distal phalanx of the 2nd finger Small epiphysis of the distal ph
## [1] "Enlarged epiphysis of the middle phalanx of the 2nd finger Small epiphysis of the middle ph
## [1] "Enlarged epiphysis of the distal phalanx of the thumb
                                                      Small epiphysis of the distal phalanx
## [1] "Enlarged thumb epiphysis Small thumb epiphysis"
## [1] "Reduced number of teeth
                           Increased number of teeth"
## [1] "Short distal phalanx of finger Long distal phalanx of finger"
## [1] "Enlarged epiphysis of the 1st metacarpal
                                           Small epiphysis of the 1st metacarpal"
## [1] "Short metacarpal Long metacarpals"
## [1] "Enlarged epiphysis of the 1st metatarsal
                                           Small epiphysis of the 1st metatarsal"
## [1] "Enlarged epiphyses of the phalanges of the hand
                                                 Small epiphyses of the phalanges of the hand
## [1] "Enlarged epiphyses of the distal phalanges of the hand Small epiphyses of the distal phalan
## [1] "Enlarged epiphyses of the middle phalanges of the hand Small epiphyses of the middle phalan
## [1] "Hyperpituitarism Hypopituitarism"
## [1] "Enlarged epiphyses Small epiphyses"
## [1] "Hypoplasia of the premaxilla
                                Hyperplasia of the premaxilla"
## [1] "Broad uvula
                 Narrow uvula"
## [1] "Long uvula
                   Short uvula"
## [1] "Increased urinary copper concentration
                                         Decreased urinary copper concentration"
## [1] "Hyperlipoproteinemia" Hypolipoproteinemia"
## [1] "Increased corneal thickness" Decreased corneal thickness"
                                                Decreased circulating luteinizing hormone le
## [1] "Elevated circulating luteinizing hormone level
## [1] "Granulocytic hyperplasia
                             Granulocytic hypoplasia"
## [1] "Monocytosis
                   Monocytopenia"
## [1] "Increased urine alpha-ketoglutarate concentration
                                                  Decreased urine alpha-ketoglutarate conce
## [1] "Hypocitraturia Hypercitraturia"
## [1] "Enlarged pituitary gland
                              Small pituitary gland"
## [1] "Hyponatriuria Hypernatriuria"
## [1] "Hypermagnesiuria
                       Hypomagnesiuria"
## [1] "Narrow jaw
                   Broad jaw"
## [1] "Increased serum testosterone level
                                      Decreased serum testosterone level"
## [1] "Increased circulating androgen level
                                        Decreased circulating androgen level"
## [1] "Chorioretinal hypopigmentation
                                   Chorioretinal hyperpigmentation"
## [1] "Long lower eyelashes Short lower eyelashes"
## [1] "Increased head circumference
                                Decreased head circumference"
## [1] "Increased size of the clitoris" Decreased size of the clitoris"
```

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## [1] "Enlarged epiphyses of the 5th toe
                                              Small epiphyses of the 5th toe"
## [1] "Enlarged epiphysis of the distal phalanx of the 2nd toe
                                                                    Small epiphysis of the distal phala
                                                                    Small epiphysis of the middle phala
## [1] "Enlarged epiphysis of the middle phalanx of the 2nd toe
## [1] "Enlarged epiphysis of the proximal phalanx of the 2nd toe
                                                                      Small epiphysis of the proximal p
## [1] "Enlarged epiphysis of the distal phalanx of the 3rd toe
                                                                    Small epiphysis of the distal phala
## [1] "Enlarged epiphysis of the middle phalanx of the 3rd toe
                                                                    Small epiphysis of the middle phala:
## [1] "Enlarged epiphysis of the proximal phalanx of the 3rd toe
                                                                      Small epiphysis of the proximal p
## [1] "Enlarged epiphysis of the distal phalanx of the 4th toe
                                                                     Small epiphysis of the distal phala
## [1] "Enlarged epiphysis of the middle phalanx of the 4th toe
                                                                     Small epiphysis of the middle phala
## [1] "Enlarged epiphysis of the proximal phalanx of the 4th toe
                                                                      Small epiphysis of the proximal p
## [1] "Enlarged epiphysis of the distal phalanx of the 5th toe
                                                                    Small epiphysis of the distal phala:
## [1] "Enlarged epiphysis of the middle phalanx of the 5th toe
                                                                    Small epiphysis of the middle phala
## [1] "Enlarged epiphysis of the proximal phalanx of the 5th toe
                                                                      Small epiphysis of the proximal p
## [1] "Long ear
                     Short ear"
```

RO exploration

I extracted the terms from the current version of the RO, then got the absolute frequencies of their words:

 $/Users/kevincohen/Dropbox/Scripts-new/lexical Frequency.pl\ experimental-outputs/ro.terms.txt$

... which reveals imbalances in the use of words that are clear opposites, e.g.:

- during 11 before 4 after 2
- to 49 from 16 towards 1 how about away?
- directly 18 indirectly 8
- indirect 2 direct 2

... which suggests that I could be adding a bunch of opposite terms to the ontology.

So, I ran the current version of my antonym-finding script:

 $code/findAntonyms.pl\ resources/ontologies/ro.2021-03-08.obo.txt\ /wc\ -l$

... which finds 43 pairs of opposites.

Next step: find overlap with Mike's resources/ontologies/predicates.txt.

For next week...

- 1. Handle substitutions (e.g. hypercalcemia vs. hypocalcemia)
- 2. Suffixes (e.g. hydrophilic vs. hydrophobic)
- 3. Word-internal, possibly (might produce a lot of false positives later when we do generation)

For reproducibility

```
## R version 4.0.1 (2020-06-06)
## Platform: x86_64-apple-darwin17.0 (64-bit)
## Running under: macOS 10.16
##
## Matrix products: default
## BLAS: /Library/Frameworks/R.framework/Versions/4.0/Resources/lib/libRblas.dylib
## LAPACK: /Library/Frameworks/R.framework/Versions/4.0/Resources/lib/libRlapack.dylib
####
```

```
## locale:
## [1] en_US.UTF-8/en_US.UTF-8/en_US.UTF-8/C/en_US.UTF-8/en_US.UTF-8
## attached base packages:
## [1] stats
                 graphics grDevices utils
                                                datasets methods
                                                                    base
##
## other attached packages:
## [1] ontologyIndex_2.7
                                                 stringr_1.4.0
                            forcats_0.5.1
##
   [4] dplyr_1.0.4
                            purrr_0.3.4
                                                 readr 1.4.0
  [7] tidyr_1.1.1
                            tibble_3.0.1
                                                 ggplot2_3.3.2
## [10] tidyverse_1.3.0
                            ggVennDiagram_0.5.0
##
## loaded via a namespace (and not attached):
## [1] Rcpp_1.0.4.6
                             lubridate_1.7.9
                                                   class_7.3-17
## [4] assertthat_0.2.1
                             digest_0.6.25
                                                   utf8_1.1.4
## [7] R6_2.4.1
                             cellranger_1.1.0
                                                   futile.options_1.0.1
## [10] backports_1.1.8
                             reprex_1.0.0
                                                   evaluate_0.14
## [13] e1071 1.7-3
                             httr 1.4.1
                                                   pillar 1.4.4
## [16] rlang_0.4.10
                             readxl_1.3.1
                                                   VennDiagram_1.6.20
## [19] rstudioapi 0.11
                             rmarkdown 2.3
                                                   labeling_0.3
## [22] munsell_0.5.0
                             broom_0.7.4
                                                   compiler_4.0.1
## [25] modelr 0.1.8
                             xfun 0.15
                                                   pkgconfig_2.0.3
## [28] htmltools_0.5.0
                             tidyselect_1.1.0
                                                   fansi_0.4.1
## [31] crayon 1.3.4
                             dbplyr_2.1.0
                                                   withr 2.4.1
## [34] sf 0.9-8
                             grid_4.0.1
                                                   jsonlite_1.7.0
                             lifecycle_0.2.0
## [37] gtable_0.3.0
                                                   DBI 1.1.0
## [40] magrittr_1.5
                             formatR_1.8
                                                   units_0.7-1
## [43] scales_1.1.1
                             KernSmooth_2.23-17
                                                   cli_2.0.2
## [46] stringi_1.4.6
                             farver_2.0.3
                                                   fs_1.4.1
## [49] xml2_1.3.2
                             futile.logger_1.4.3
                                                   ellipsis_0.3.1
## [52] generics_0.0.2
                             vctrs_0.3.6
                                                   lambda.r_1.2.4
## [55] tools_4.0.1
                             glue_1.4.1
                                                   hms_1.0.0
## [58] yaml_2.2.1
                             colorspace_1.4-1
                                                   classInt_0.4-3
## [61] rvest_0.3.6
                             knitr_1.29
                                                   haven_2.3.1
```