# Lab notebook

### **KBC**

## 4/5/2021

#### 2021-04-05

I extracted the terms from the current version of the RO, then got the absolute frequencies of their words: /Users/kevincohen/Dropbox/Scripts-new/lexicalFrequency.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
- 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:

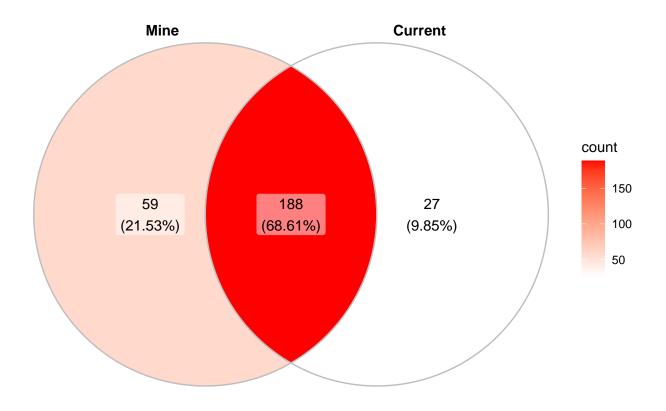
 $code/findAntonyms.pl\ resources/ontologies/ro.2021-03-08.obo.txt\ /wc\ -l$ 

... which finds 43 pairs of opposites.

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

### PATO: opposites already annotated

```
## 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()
# NB: need to add reduce/increase. Do HP, then RO.
# only found by me
# comm -23 experimental-outputs/pato.mine.terms.only.tsv resources/current-opposites.sorted.terms.only.
      59
# only in current annotations
#comm -13 experimental-outputs/pato.mine.terms.only.tsv resources/current-opposites.sorted.terms.only.t
      27
# in both
#comm -12 experimental-outputs/pato.mine.terms.only.tsv resources/current-opposites.sorted.terms.only.t
     188
# https://www.datanovia.com/en/fr/blog/magnifique-diagramme-de-venn-ggplot-avec-r/
# this separates out the three entirely--not what the ggVennDiagram() function is looking for
# Here I'm just getting the terms themselves in printable fashion, separating them by who "finds" them
pato.mine.only <- as.vector(read_lines("/Users/kevincohen/Dropbox/N-Z/translator-concept-oppositeness/e
pato.mine.only <- stringr::str_split(pato.mine.only, pattern = "\t")</pre>
current.only <- as.vector((read_lines("/Users/kevincohen/Dropbox/N-Z/translator-concept-oppositeness/ex</pre>
current.only <- stringr::str_split(current.only, pattern = "\t")</pre>
both.mine.and.current <- as.vector((read_lines("/Users/kevincohen/Dropbox/N-Z/translator-concept-opposi
both.mine.and.current <- stringr::str_split(both.mine.and.current, pattern = "\t")
# here I have *everything* that I found and *everything* in current, so that we can quantify overlap &
pato.mine <- as.vector(read_lines("/Users/kevincohen/Dropbox/N-Z/translator-concept-oppositeness/expering)</pre>
current <- as.vector(read_lines("/Users/kevincohen/Dropbox/N-Z/translator-concept-oppositeness/resource</pre>
x <- list(Mine = pato.mine, Current = current)</pre>
ggVennDiagram(x)
```

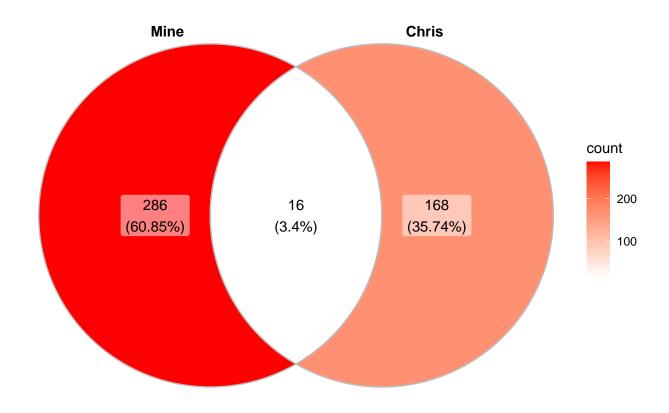


```
# print("In both mine and 'current':")
# print(both.mine.and.current, row.names = FALSE)
# print("In mine only:")
# print(pato.mine.only)
# print("In 'current' only:")
# print(current.only)
#as.tibble(pato.mine.only)
```

# Comparison to Chris et al.'s stuff

```
chris <- read.table("/Users/kevincohen/Dropbox/N-Z/translator-concept-oppositeness/experimental-outputs
colnames(chris) <- c("term01", "term02")
chris <- as_tibble(chris)
hp.mine <- read.table("/Users/kevincohen/Dropbox/N-Z/translator-concept-oppositeness/experimental-output
colnames(hp.mine) <- c("term01", "term02")
hp.mine <- as_tibble(hp.mine)

# Reran my stuff on HPO 2021-04
chris.as.vector <- as.vector(paste(chris$term01, chris$term02))
hp.mine.as.vector <- as.vector(paste(hp.mine$term01, hp.mine$term02))
x <- list(Mine = hp.mine.as.vector, Chris = chris.as.vector)
ggVennDiagram(x)</pre>
```



```
print("Mine:")
## [1] "Mine:"
print(hp.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
print("Chris's:")
```

## [1] "Chris's:"

```
print(chris)
## # 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.
library(ontologyIndex)
## Warning: package 'ontologyIndex' was built under R version 4.0.2
# His data is IDs only, and we need the terms, so we need to have a searchable version of HPO.
hpo.ontology <- get_ontology("/Users/kevincohen/Dropbox/a-m/Corpora/obo/hp-2020-20-12.obo")
for (i in 1:nrow(chris)) {
  #print(chris[i,])
  opposites <- chris[i,]
  id01 <- as.character(opposites[1])</pre>
  id02 <- as.character(opposites[2])</pre>
  term01 <- get_term_property(hpo.ontology, property = "name", term = id01)</pre>
  term02 <- get_term_property(hpo.ontology, property = "name", term = id02)
  print(paste(term01, term02, sep = "
  # term01 <- chris[i,]
  # term02 <- chris[i,]
}
## [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"
                      Narrow nose"
## [1] "Wide nose
## [1] "Short neck
                       Long neck"
## [1] "Microcornea
                        Megalocornea"
## [1] "Long eyelashes
                           Short eyelashes"
## [1] "Thick eyebrow
                          Thin eyebrow"
                                    Short palpebral fissure"
## [1] "Long palpebral fissure
                       Increased number of teeth"
## [1] "Hypodontia
## [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"
## [1] "Long hallux
                        Short hallux"
## [1] "Leukopenia
                       Leukocytosis"
## [1] "Hypoglycemia
                         Hyperglycemia"
## [1] "Hyperammonemia
                           Hypoammonemia"
## [1] "Hypophosphatemia
                             Hyperphosphatemia"
## [1] "Hyperuricemia
                          Hypouricemia"
## [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] "Large sella turcica
                                Small sella turcica"
## [1] "Lymphadenopathy
                            Lymph node hypoplasia"
## [1] "Decreased lymphocyte apoptosis
                                            Increased lymphocyte apoptosis"
## [1] "Hypoventilation
                            Hyperventilation"
                                             Increased circulating IgM level"
## [1] "Decreased circulating total IgM
## [1] "Hypocalcemia
                         Hypercalcemia"
## [1] "Hyponatremia
                         Hypernatremia"
## [1] "Hyperchloriduria
                             Hypochloriduria"
## [1] "Hypomagnesemia
                           Hypermagnesemia"
## [1] "Decreased circulating ACTH level
                                              Increased circulating ACTH level"
## [1] "Hypoalbuminemia
                            Hyperalbuminemia"
```

Hypolipidemia"

## [1] "Hyperlipidemia

```
## [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"
                                          Low alkaline phosphatase"
## [1] "Elevated alkaline phosphatase
## [1] "Decreased HDL cholesterol concentration
                                                    Increased HDL cholesterol concentration"
## [1] "Hypermethioninemia
                               Hypomethioninemia"
## [1] "Increased circulating IgG level
                                            Decreased circulating IgG level"
## [1] "Elevated serum creatinine
                                     Decreased serum creatinine"
## [1] "Increased serum ferritin
                                     Decreased serum ferritin"
## [1] "Decreased urinary sulfate
                                     Increased urinary sulfate"
## [1] "CNS hypomyelination
                               CNS hypermyelination"
## [1] "Large humeral epiphyses Small humeral epiphyses"
## [1] "Large radial epiphyses
                                   Small radial epiphyses"
## [1] "Short palm
                      Long palm"
## [1] "Decreased circulating antibody level
                                                 Increased circulating antibody level"
                                  Decreased body weight"
## [1] "Increased 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] "Decreased mean platelet volume
                                          Increased mean platelet volume"
## [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"
## [1] "Large basal ganglia
                               Small basal ganglia"
## [1] "Peripheral hypomyelination Peripheral hypermyelination"
## [1] "Generalized hyperpigmentation Generalized hypopigmentation"
## [1] "Long upper eyelashes
                                 Short upper eyelashes"
## [1] "Macular hypopigmentation Macular hyperpigmentation"
## [1] "Foveal hyperpigmentation
                                   Foveal hypopigmentation"
## [1] "Decreased activity of mitochondrial respiratory chain
                                                                  Increased activity of mitochondrial r
## [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] "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 epiphysis of the proximal phalanx of the 3rd finger
                                                                          Small epiphysis of the proxima
## [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
## [1] "Enlarged epiphyses of the 2nd finger"
Small 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 proximal phalanx of the thumb

## [1] "Enlarged epiphysis of the proximal phalanx of the 2nd finger Small epiphysis of the proxima

Small epiphysis of the proximal pha

```
## [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 epiphyses of the hallux
                                             Small epiphyses of the hallux"
## [1] "Enlarged epiphysis of the proximal phalanx of the hallux
                                                                     Small epiphysis of the proximal ph
## [1] "Enlarged epiphysis of the distal phalanx of the hallux
                                                                   Small epiphysis of the distal phalan
## [1] "Enlarged epiphysis of the 1st metatarsal
                                                     Small epiphysis of the 1st metatarsal"
## [1] "Enlarged epiphyses of the toes
                                           Small epiphyses of the toes"
## [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] "Enlarged epiphyses of the proximal phalanges of the hand
                                                                   Small epiphyses of the proximal ph
## [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"
## [1] "Decreased urinary urate
                                    Increased urinary urate"
## [1] "Elevated circulating luteinizing hormone level
                                                           Decreased circulating luteinizing hormone le
## [1] "Erythroid hyperplasia
                                  Erythroid hypoplasia"
## [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] "Decreased inflammatory response
                                            Increased inflammatory response"
## [1] "Decreased pineal volume
                                    Increased pineal volume"
## [1] "Narrow jaw
                      Broad jaw"
## [1] "Increased serum testosterone level
                                               Decreased serum testosterone level"
## [1] "Increased circulating androgen level
                                                Decreased circulating androgen level"
## [1] "Decreased mitochondrial number
                                           Increased mitochondrial number"
## [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"
## [1] "Decreased size of nasopharyngeal adenoids
                                                      Increased size of nasopharyngeal adenoids"
## [1] "Enlarged epiphyses of the 2nd toe
                                              Small epiphyses of the 2nd toe"
## [1] "Enlarged epiphyses of the 3rd toe
                                              Small epiphyses of the 3rd toe"
                                             Small epiphyses of the 4th toe"
## [1] "Enlarged epiphyses of the 4th toe
## [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
## [1] "Enlarged epiphysis of the middle phalanx of the 2nd toe
                                                                    Small epiphysis of the middle phala
## [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
```

Small epiphysis of the middle phala

Small epiphysis of the proximal p

## [1] "Enlarged epiphysis of the middle phalanx of the 3rd toe

## [1] "Enlarged epiphysis of the proximal phalanx of the 3rd toe

```
## [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"
intersection.ids <- intersect(hp.mine, chris)</pre>
if (DEBUG) { print(intersection.ids) }
for (i in 1:nrow(intersection.ids)) {
  opposites <- intersection.ids[i,]
  id01 <- as.character(opposites[1])</pre>
  id02 <- as.character(opposites[2])</pre>
  term01 <- get_term_property(hpo.ontology, property = "name", term = id01)</pre>
  term02 <- get_term_property(hpo.ontology, property = "name", term = id02)</pre>
                                          "))
  print(paste(term01, term02, sep = "
## [1] "Decreased activity of mitochondrial respiratory chain
                                                                   Increased activity of mitochondrial r
## [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 lymphocyte apoptosis
                                            Increased lymphocyte apoptosis"
## [1] "Decreased mean platelet volume
                                            Increased mean platelet volume"
## [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"
                                Small sella turcica"
## [1] "Large sella turcica
mine.only.ids <- setdiff(hp.mine, chris)
chris.only.ids <- setdiff(chris, hp.mine)</pre>
# TODO: could make term-only tibbles here
for (i in 1:nrow(mine.only.ids)) {
  opposites <- mine.only.ids[i,]
  id01 <- as.character(opposites[1])</pre>
  id02 <- as.character(opposites[2])</pre>
  term01 <- get_term_property(hpo.ontology, property = "name", term = id01)</pre>
  term02 <- get_term_property(hpo.ontology, property = "name", term = id02)</pre>
  print(paste(term01, term02, sep = "
## [1] "Abnormal dark-adapted electroretinogram
                                                     Abnormal light-adapted electroretinogram"
## [1] "Abnormal hard palate morphology Abnormal soft palate morphology"
```

## [1] "Abnormal large intestinal mucosa morphology" Abnormal small intestinal mucosa morphology"

```
## [1] "Acellular urinary casts
                                  Cellular urinary casts"
## [1] "Activating thyroid-stimulating hormone receptor defect
                                                                   Inactivating thyroid-stimulating hor
## [1] "Acute bronchitis
                             Chronic bronchitis"
## [1] "Acute colitis
                          Chronic colitis"
                               Chronic constipation"
## [1] "Acute constipation
                                  Chronic cutaneous wound"
## [1] "Acute 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"
                                                Symmetric peripheral demyelination"
## [1] "Asymmetric peripheral demyelination
## [1] "Atonic seizure
                           Tonic seizure"
                                     Typical absence seizure"
## [1] "Atypical absence seizure
## [1] "Atypical absence status epilepticus
                                                Typical absence status epilepticus"
## [1] "Cleft hard palate
                             Cleft soft palate"
## [1] "Closed comedo
                          Open 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
                                                                    Partial duplication of the phalange
## [1] "Complete duplication of the phalanges of the 5th finger
## [1] "Complete duplication of the proximal phalanges of the hand
                                                                       Partial duplication of the proximation
## [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
                                                                           Partial duplication of the p
## [1] "Complete duplication of the proximal phalanx of the 3rd finger
## [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 p
## [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"
                                        Increased corneal thickness"
## [1] "Decreased corneal thickness
## [1] "Decreased CSF alanine concentration
                                                Increased CSF alanine concentration"
## [1] "Decreased CSF albumin concentration
                                                Increased CSF albumin concentration"
## [1] "Decreased CSF arginine concentration
                                                 Increased CSF arginine concentration"
## [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"
                                                      Increased CSF phenylalanine concentration"
  [1] "Decreased CSF phenylalanine concentration
## [1] "Decreased CSF protein
                                  Increased CSF protein"
## [1] "Decreased CSF serine concentration
                                               Increased 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 glomerular filtration rate
                                                Increased glomerular filtration rate"
## [1] "Decreased glucagon level
                                     Increased glucagon level"
## [1] "Decreased glucose-6-phosphate dehydrogenase level in blood
                                                                       Increased glucose-6-phosphate del
## [1] "Decreased glucose-6-phosphate dehydrogenase level in dried blood spot
                                                                                  Increased glucose-6-p
## [1] "Decreased glucose-6-phosphate dehydrogenase level in leukocytes
                                                                             Increased glucose-6-phospha
## [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"
## [1] "Decreased level of platelet-activating factor
                                                      Increased 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 mean corpuscular volume Increased mean corpuscular volume"
## [1] "Decreased monosialylated core 1 O-glycan level
                                                      Increased monosialylated core 1 O-glycan lev
## [1] "Decreased muscle glycogen content" Increased muscle glycogen content"
## [1] "Decreased phosphoribosylpyrophosphate synthetase level Increased phosphoribosylpyrophosphat
## [1] "Decreased pituitary glycoprotein hormone alpha subunit level Increased pituitary glycoprote
## [1] "Decreased proportion of CD4+CD25+ regulatory T cells
                                                            Increased proportion of CD4+CD25+ regu
## [1] "Decreased proportion of CD4-positive, alpha-beta memory T cells Increased proportion of CD4
## [1] "Decreased proportion of CD8-positive, alpha-beta memory T cells Increased proportion of CD8
## [1] "Decreased proportion of CD8-positive, alpha-beta TEMRA T cells Increased proportion of CD8-
## [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
## [1] "Decreased proportion of memory B cells
                                             Increased proportion of memory B cells"
## [1] "Decreased proportion of memory T cells
                                                Increased 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 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 salivary cortisol level
                                           Increased salivary cortisol level"
## [1] "Decreased scrotal rugation
                                   Increased scrotal rugation"
## [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"
                                                     Increased serum insulin-like growth factor 1"
## [1] "Decreased 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 skull ossification"

## [1] "Decreased skull ossification

```
## [1] "Decreased T3/T4 ratio
                        Increased 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"
## [1] "Decreased urine alpha-ketoglutarate concentration
                                                Increased urine alpha-ketoglutarate conce
## [1] "Decreased VLDL cholesterol concentration
                                         Increased VLDL cholesterol concentration"
## [1] "Decreased waist to hip ratio" Increased waist to hip ratio"
## [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"
                             Small cell lung carcinoma"
## [1] "Large 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] "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] "Nonproductive cough Productive cough"

## [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"

```
Progressive cerebellar ataxia"
## [1] "Nonprogressive cerebellar ataxia
## [1] "Nonprogressive encephalopathy
                                          Progressive encephalopathy"
## [1] "Nonprogressive visual loss
                                       Progressive visual loss"
                           Progressive"
## [1] "Nonprogressive
## [1] "obsolete Decreased proportion of CD4+ central memory cells
                                                                        obsolete Increased proportion of
## [1] "Primary amenorrhea
                               Secondary amenorrhea"
## [1] "Primary Caesarian section
                                      Secondary Caesarian section"
## [1] "Primary hyperaldosteronism
                                       Secondary hyperaldosteronism"
## [1] "Primary hypercortisolism
                                     Secondary hypercortisolism"
## [1] "Primary hyperparathyroidism
                                        Secondary hyperparathyroidism"
## [1] "Secondary hyperparathyroidism
                                          Tertiary hyperparathyroidism"
## [1] "Submucous cleft hard palate
                                        Submucous cleft soft palate"
## [1] "Undetectable dark-adapted electroretinogram
                                                        Undetectable light-adapted electroretinogram"
for (i in 1:nrow(chris.only.ids)) {
  opposites <- chris.only.ids[i,]</pre>
  id01 <- as.character(opposites[1])</pre>
  id02 <- as.character(opposites[2])</pre>
  term01 <- get_term_property(hpo.ontology, property = "name", term = id01)</pre>
  term02 <- get_term_property(hpo.ontology, property = "name", term = id02)</pre>
  print(paste(term01, term02, sep = "
## [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"
                              Small fontanelle"
## [1] "Large fontanelles
## [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"
                           Thin nail"
## [1] "Onychogryposis
## [1] "Broad nail
                       Narrow nail"
## [1] "Short toe
                      Long toe"
## [1] "Long hallux
                        Short hallux"
## [1] "Leukopenia
                       Leukocytosis"
## [1] "Hypoglycemia
                         Hyperglycemia"
## [1] "Hyperammonemia
                           Hypoammonemia"
## [1] "Hypophosphatemia
                             Hyperphosphatemia"
## [1] "Hyperuricemia
                          Hypouricemia"
## [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"
## [1] "Increased circulating IgG level
                                            Decreased circulating IgG level"
## [1] "Elevated serum creatinine
                                      Decreased serum creatinine"
## [1] "Increased serum ferritin
                                     Decreased serum ferritin"
                                CNS hypermyelination"
## [1] "CNS hypomyelination
## [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"
## [1] "Peripheral hypomyelination
                                     Peripheral hypermyelination"
## [1] "Generalized hyperpigmentation
                                        Generalized hypopigmentation"
                               Short upper eyelashes"
## [1] "Long 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] "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 epiphysis of the proximal phalanx of the 3rd finger
                                                                      Small epiphysis of the proxima
## [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 proximal phalanx of the 2nd finger Small epiphysis of the proxima
## [1] "Enlarged epiphysis of the proximal phalanx of the thumb
                                                               Small epiphysis of the proximal pha
## [1] "Enlarged epiphysis of the distal phalanx of the thumb
                                                               Small epiphysis of the distal phalanx
                                  Small thumb epiphysis"
## [1] "Enlarged 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 epiphyses of the hallux
                                           Small epiphyses of the hallux"
## [1] "Enlarged epiphysis of the distal phalanx of the hallux
                                                                Small epiphysis of the distal phalan
## [1] "Enlarged epiphysis of the 1st metatarsal
                                                   Small epiphysis of the 1st metatarsal"
## [1] "Enlarged epiphyses of the toes
                                         Small epiphyses of the toes"
## [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
                                                                Small epiphyses of the middle phalan
## [1] "Enlarged epiphyses of the middle phalanges of the hand
## [1] "Enlarged epiphyses of the proximal phalanges of the hand
                                                                 Small epiphyses of the proximal ph
## [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"
## [1] "Elevated circulating luteinizing hormone level
                                                        Decreased circulating luteinizing hormone le
## [1] "Erythroid hyperplasia
                              Erythroid hypoplasia"
## [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"
                                               Decreased serum testosterone level"
## [1] "Increased 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"
                                              Small epiphyses of the 2nd toe"
## [1] "Enlarged epiphyses of the 2nd toe
## [1] "Enlarged epiphyses of the 3rd toe
                                              Small epiphyses of the 3rd toe"
## [1] "Enlarged epiphyses of the 4th toe
                                              Small epiphyses of the 4th toe"
## [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
## [1] "Enlarged epiphysis of the middle phalanx of the 2nd toe
                                                                    Small epiphysis of the middle phala
## [1] "Enlarged epiphysis of the proximal phalanx of the 2nd toe
                                                                      Small epiphysis of the proximal p
                                                                    Small epiphysis of the distal phala
## [1] "Enlarged epiphysis of the distal phalanx of the 3rd toe
## [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"
```