

# Data Dictionary And Documentation

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#### **Introduction**

This document describes the analysis datasets and derived variables for the Long Life Family Study (Visit 1 and Visit 2). In general, each form/procedure in either Pre-Clinic (PTSI, Relatives), Clinic or Follow-Up is stored as its own SAS dataset (e.g. blood, meds, physical, venip, etc.), with multiple records (observation) per person corresponding to what visit the data was collected during (see below). The few exceptions to this rule are noted in the appropriate places. For the most part, each dataset retains the original, raw form variables as collected on each subject. These are mnemonically named (e.g. SEX instead of Q7), with accompanying SAS labels. A user-defined format library is also included to provide value-labels to codes. Thus, PROC CONTENTS, along with PROC FORMAT with the FMTLIB option can be used in conjunction with the official book of forms and QxQs (also supplied by the Coordinating Center) to provide documentation for the raw form data itself. We concentrate here instead on documenting the dataset organization, and derived analysis variables created at the Coordinating Center.

The data sets for each Panel/Form may contain multiple records/rows per subject with each row corresponding to a different exam/follow-up (depending on how many times the Form was administered on the subject). The variables Visitcode and/or Contactyr in a data set help us to track down which event the data was collected from. The label and values for Visitcode and Contactyr are:

#### Visitcode (Visit code):

1 - Visit 1 in-person
2 - Visit 1 Follow-up
3 - Visit 2 in-person (returning participants)
4 - Visit 2 in-person (new participants)
5 - Visit 2 follow-up
Contactyr (follow-up contact year):
0 - Visit 1 in-person
1, 2, 3, 4 ..., or 9 - follow-up year 1, 2, 3, 4 ..., or 9
99 - Visit 2 in-person

#### **Derived Variables**

For the most part, all derived variables are named beginning with an underscore, to readily distinguish them from the raw form variables. The derived variables are stored in the "natural" form/procedure dataset from which they were created, e.g. \_BMI is in BPHR dataset, total cholesterol is in the BLOOD dataset, etc. In the various sections below, we summarize each dataset, followed by a description of each derived variable in it. In many cases, we give not only the algorithm used in defining the variable, but also the actual SAS code which implements it, for completeness, and to assure that the final coding is as intended. Each of the derived variables is described first, then the SAS code that created all of them is presented.

#### **Common Variables**

Most datasets have a few, common identifying variables.

#### ID

ID is the original key identifying variable, at the time of data collection. ID is usually in each dataset. It is the 8 character LLFS ID. The structure of ID is:

CNNNNNNN C is the single digit field center number: 1 = Denmark, DK 2 = Boston, MA 3 = Pittsburgh, PA 4 = New York, NY

#### TOUCHDAT

TOUCHDAT is a SAS datetime variable automatically updated by the Data Entry System at Field Centers to give the date/time when the record was last changed.

#### DATE

DATE is a SAS date variable giving the date the form/procedure was collected.

#### FC

FC is the field center. 1 = Denmark, DK 2 = Boston, MA 3 = Pittsburgh, PA 4 = New York, NY **SUBJECT** 

SUBJECT is a de-identified, unique identifier for each Participant. It is a 5 digit number. Datasets with one obs/subject are uniquely identified by ID and can be merged/linked using this variable.

## PHASE I

## Pre-In Person Visit

## Family Longevity Collection Score Instrument – Proband (FLoSS) (PTSI) Dataset

This dataset contains the answers to eligibility questions, including those that were used to calculate the Family Longevity Selection Score (FLoSS), and some socio-demographic data. This dataset has no derived variables.

## Proband Relative Contact Information (Relatives) Dataset

This questionnaire collected information on the Proband's relatives, such as contact information, and permission to contact them.

This dataset has no derived variables.

### Sibling Information (SIBINFO) Dataset

This dataset contains information on the Proband's siblings. This dataset has no derived variables.

## In Person Visit

## Blood (Blood) Dataset

This dataset contains the data regarding the blood chemistries, the date the blood was drawn, and the LLFS Subject. Special assays and candidate genes were added as well. This dataset has four derived variables:

#### \_FTESTSTRN - Free Testosterone (ng/dL)

Free T (mol/L) =  $\frac{-b+\sqrt{b^2+4a[TT]}}{2a}$ , where a = ka + ks + (ka × ks) ([SHBG (mol/L)] + [Alb (mol/L)] - [TT (mol/L)]), b = 1 + ks [SHBG] + ka [Alb] - (ka + ks) [TT], ka = 3.6×10<sup>4</sup> L/mol, ks =1×10<sup>9</sup> L/mol. Total testosterone (TT) (mol/L) = (Reported TT (ng/dl)/288.4) x 10<sup>-8</sup> Molecular weight of Testosterone = 288.4 SHBG (mol/L) = Reported SHBG (nmol/L) x 10<sup>-9</sup> Alb (mol/L) = (Reported Alb (g/dl) x 10)/69000 Molecular weight of Albumin = 69000

\_ALBBT - Albumin Bound Testosterone (ng/dL)

Albumin Bound T (ng/dl) = [ka x (C<sub>onc.</sub>Alb(mol/L)]\*Free T where ka =  $3.6 \times 10^4$  L/mol

\_BIOTESTSTRN - Bioavailable Testosterone (ng/dL)

Bioavailable Testosterone = Free Testosterone + Albumin bound Testosterone

\_GLUR\_NEW that replaces GLUR (glucose) with some updates.

For the subjects whose blood samples were processed after two hours of collection, their lab measured glucose levels were likely underestimated because of glycolysis (sugar breaking down) at room temperature. However, in most of nondiabetic subjects among them, glucose levels may be best estimated through their HbA1c levels (estimated glucose or eAG levels = 28.7\*HbA1c - 46.7, Nathan DM et al. Diabetes Care 2008;31:1-6 followed by some necessary

adjustments). In specific, glucose levels = alpha \* eAG in nondiabetic subjects whose blood samples were processed within two hours of collection. And thereafter, for each nondiabetic subject whose sample was processed at least two hours after collection of blood, alpha was applied to obtain his/her best estimated glucose value (glucose = alpha \* eAG). The estimation of alpha is performed for visit 1 and visit 2 separately. Please note that the visit 2 data subject to change based on final data. The SAS code that creates \_GLUR\_NEW is as follows:

```
if drawtime>0 and t26time >0 then processtime = INTCK('minute',drawtime,t26time)/60;
  label drawtime = 'Time Venipuncture ended'
       t26time = 'Time SST1 & SST2 tubes 2&6 were spun';
if processtime>=0 and processtime<=2 then gt2hrs=0;
else if processtime>2 then gt2hrs=1;
else if processtime<0 then gt2hrs=-1;
eAG = glyhb * 28.7 - 46.7;
   label eAG='Estimated Average Glucose (mg/dL)';
if (fast=1 and glur>=126) or glyhb >=6.5 or diabnow=1 or diab=1 then diabetes=1;
else diabetes=0;
alpha = glur_mean / eag_mean;
** glur mean is mean of glur and eag mean is mean of eAG. They are
estimated using fasting non-T2D samples that were processed within two hours of
collection. The estimations are performed for Visit 1 and Visit 2 separately;
if _diabetes=1 then glur_new = .;
if gt2hrs=1 and diabetes=0 then glur new = &alpha0 * eag;
if (gt2hrs=0 or gt2hrs=-1) and diabetes=0 then glur new = glur;
```

## Blood Pressure, Heart Rate, Height, Weight, Waist (BPHR) Dataset

This dataset contains the data regarding the blood pressure, height, knee height and waist circumference.

#### \_HEIGHT

\_HEIGHT is the Average Standing Height in cm calculated from stand1, stand2, stand3, and stand4.

```
if stand1>0 and stand2>0 and (not(stand3>0) or not(stand4>0))
        then _height=mean(stand1,stand2);
else if stand3>0 and stand4>0
        then height=mean(stand3,stand4);
```

#### \_BMI

\_BMI is Body Mass Index calculated from weight and height on BPHR.

```
if (weight >0 and _height>0) then
    BMI=(weight/(( height/100)**2));
```

#### \_WAIST

\_WAIST is the Average Abdominal Circumference in cm calculated from waist1, waist2, waist3, and waist4.

```
if waist1>0 and waist2>0 and (not(waist3>0) or not(waist4>0))
    then _waist=mean(waist1,waist2);
    else if waist3>0 and waist4>0
        then _waist=mean(waist3,waist4);
```

#### \_SIT

\_SIT is the Average Height while sitting in cm calculated from sit1, sit2, sit3, and sit4.

```
if sit1>0 and sit2>0 and (not(sit3>0) or not(sit4>0))
        then _sit=mean(sit1,sit2);
else if sit3>0 and sit4>0
        then _sit=mean(sit3,sit4);
```

#### \_HTIN25 & \_HTCM25

\_HTIN25 is the Height when 25 years old in total inches. \_HTCM25 is the Height when 25 years old in cm.

```
if htft25>0 and htin25>=0 then _htin25= (htft25 * 12) + htin25;
if htft25>0 and htin25<0 then _htin25= (htft25 * 12);
if htcm25>0 then _htin25= htcm25 / 2.54;
_htcm25 = _htin25 * 2.54;
```

#### \_KNEE

\_KNEE is the Average Length of Leg from Heel to Knee in cm.

\_knee=mean(knee1,knee2);

#### \_SBP

\_SBP is the Average Sitting Systolic Blood Pressure in mmHg.

\_sbp=mean(sys1,sys2,sys3);

#### \_DBP

\_DBP is the Average Sitting Diastolic Blood Pressure in mmHg.

\_dbp=mean(dia1,dia2,dia3);

#### \_PULSE

\_PULSE is the Average Sitting Pulse.

```
_pulse=mean(pulse1,pulse2,pulse3);
```

## Carotid Intima-Media Thickness Test Worksheet (CAROTID) Dataset

This dataset contains information from URL Carotid IMT Worksheet entered into Redcap by the sites. Its main use has been by the URL lab to check alerts, plaque, and tracking. Carotid plaque variables from this dataset are **NOT** to be used for analyses. Please use the plaque data from the **finalcqi** dataset

This dataset has no derived variables.

## <u>Carotid Intima-Media Thickness Test Function (CAROTIDFUNC)</u> <u>Dataset</u>

This dataset contains the carotid artery analysis values that were read at the Ultrasound Reading Lab at Pittsburgh. These variables should be used for carotid function analyses. Carotid assessment, primary variables for analysis, data cleaning, data collection and quality control are in the document "LLFS Carotid Data DictionaryFINAL\_ebm.pdf". This dataset has no derived variables.

## Mood and Personality Assessment (CES-D and NEO 5-Factor) Dataset

This dataset contains the data regarding depressive symptomatology, personality dimensions of neuroticism and conscientiousness. The 24 questions NEO is only administered at in person visit 1. Derived variables NEUROTICISM, and CONSCIENTIOUSNESS are sums of different questions on the form. The SIZE\_NEUROTOCOSM and SIZE\_CONSCIENTIOUSNESS are the number of non-missing responses for NEUROTOCOSM and CONSCIENTIOUSNESS, respectively, which are used to calculate the scores.

# NEUROTICISM, SIZE\_NEUROTOCOSM, CONSCIENTIOUSNESS and SIZE\_CONSCIENTIOUSNESS

```
#R scripts (from Boston field site)
cesdneo.orig.1to5.scale <- read.csv(paste(root.dir,"cesdneoall.csv",sep=""),
header=T, na.strings=c("", "D","R", "N", "U", NA) )</pre>
```

```
index.nonreverse <- c("anxious", "sad", "worrier", "blue", "wastetime", "reliable",</pre>
"organized", "methodical")
nonreverse <- cesdneo.orig.1to5.scale[, index.nonreverse]</pre>
new.nonreverse <- 5 - nonreverse</pre>
index.need.reverse <- c("acmplshgoals", "commitment", "neat", "pacing", "productive",
"conscientious", "excellence", "cleargoals", "tense", "angry", '
"worthless", "discouraged", "inferior", "stress", "helpless", "ashamed")
need.reverse <- cesdneo.orig.1to5.scale[, index.need.reverse]</pre>
reversed <- need.reverse - 1
other.index <- c("subject", "fc", "version", "date", "form_completed",
"date completed")
other <- cesdneo.orig.1to5.scale[, other.index]</pre>
length(index.nonreverse)+ length(index.need.reverse) # 24
# ======= form new data =========
cesdneo.rescaled <- data.frame(other, reversed, new.nonreverse)</pre>
cesdneo.Neuroticism.data <- data.frame(cesdneo.rescaled$tense,</pre>
cesdneo.rescaled$worthless, cesdneo.rescaled$anxious, cesdneo.rescaled$angry,
cesdneo.rescaled$discouraged, cesdneo.rescaled$sad, cesdneo.rescaled$worrier,
cesdneo.rescaled$inferior, cesdneo.rescaled$stress, cesdneo.rescaled$helpless,
cesdneo.rescaled$ashamed, cesdneo.rescaled$blue)
   names(cesdneo.Neuroticism.data) <- c("tense", "worthless", "anxious", "angry",</pre>
"discouraged", "sad", "worrier", "inferior", "stress", "helpless", "ashamed", "blue")
   length(names(cesdneo.Neuroticism.data))
cesdneo.Conscientiousness.data <- data.frame(cbind(cesdneo.rescaled$wastetime,</pre>
cesdneo.rescaled$acmplshgoals, cesdneo.rescaled$commitment,
cesdneo.rescaled$reliable, cesdneo.rescaled$neat, cesdneo.rescaled$pacing,
cesdneo.rescaled$productive, cesdneo.rescaled$organized, cesdneo.rescaled$methodical,
cesdneo.rescaled$conscientious, cesdneo.rescaled$excellence,
cesdneo.rescaled$cleargoals))
   names(cesdneo.Conscientiousness.data) <-</pre>
c("wastetime","acmplshgoals","commitment","reliable","neat","pacing", "productive",
"organized", "methodical", "conscientious", "excellence", "cleargoals")
      Neuroticism <- apply(cesdneo.Neuroticism.data, 1, sum, na.rm=T)</pre>
Conscientiousness <- apply(cesdneo.Conscientiousness.data, 1, sum, na.rm=T)
      size.Neuroticism <- apply( is.na(cesdneo.Neuroticism.data)==F,1,sum)</pre>
size.Conscientiousness <- apply( is.na(cesdneo.Conscientiousness.data)==F,1,sum)</pre>
cesdneo.rescaled <- data.frame(cesdneo.rescaled, Neuroticism, size.Neuroticism,</pre>
Conscientiousness, size.Conscientiousness)
cesdneo.rescaled <- data.frame(cesdneo.rescaled, Neuroticism, size.Neuroticism,</pre>
Conscientiousness, size.Conscientiousness)
```

## Coded Medications (CODEDMEDS) Dataset

This dataset contains the four yes/no variables Anne Newman and her staff at the University of Pittsburgh developed from the Medications data. Specifically, they are HTNRX, LIPIDRX, NITRORX, DMRX. These represent whether or not the Participant is currently taking a medication for Hypertension, Lipid Lowering, Angina, or Diabetes Mellitus. This dataset has no derived variables.

This is for visit 1 data only.

## Coded Medications (CODEDMEDS\_ATC) Dataset

This dataset contains the four yes/no variables Paola Sebastiani and her staff at the Boston University developed from the Visit 1 Medications data, using the World Health Organization's Anatomical Therapeutic Chemical (ATC) classifications. DMCC recreate 4 of them for Visit 2 Medication data based on the same coding scheme used for Visit 1. Specifically, the four variables are HTN, LIPID, NITRO, DIAB. These represent whether or not the Participant at Visit 1 or Visit 2 is taking a medication for Hypertension, Lipid Lowering, Angina, or Diabetes Mellitus.

This dataset has no derived variables. This is for Visit 1 data and Visit 2 data.

## Cognitive Assessments (COGASSESS) Dataset

There are three forms that compose this battery, the NACC UDS, the Telephone Interview for Cognitive Status (TICS, see below), and the Informant-Based Date of Onset Interview. This dataset has no derived variables.

## Consent Tracking and Interview Feasibility (CTIF) Dataset

This dataset contains the answers to the consent questions. There were different questions at the different Field Centers, based upon what their individual Internal Review Boards required. This dataset has no derived variables.

## Digital Clock Drawing Test (dCDT)Dataset

This dataset contains the variables obtained from the use of the digital pen in the clock drawing test as part of the cognitive assessment. Scored clocks were sent to the reading lab at MIT and the final data was sent to the DMCC from MIT for release. The long label for each variable is in a separate file called "eclock\_variables\_01132016.xlsx". This dataset has no derived variables

## <u>Carotid Intima-Media Thickness Test Plaque Assessment (FINALCQI)</u> <u>Dataset</u>

This dataset contains data from the Carotid Duplex Scan Feedback Form that is completed by reading center sonographers after assessing images and video clips for plaque presence and burden as well as image quality scoring. This dataset contains the carotid plaque data to be used for plaque assessment. Details for variables, data collection, and quality control are in the document "LLFS Carotid Data DictionaryFINAL\_ebm.pdf".

## Lung Function (LUNGFUNC) Dataset

This dataset contains the results of the Spirometry test, sent to the Data Coordinating Center from the Reading Center. This dataset has no derived variables.

## Medication Inventory (MEDCHK) Dataset

This dataset contains the response of the first question of the Medication Inventory; if any medication was taken in the past 2 weeks. This dataset has no derived variables.

## Medication Inventory (MEDS) Dataset

This dataset contains the responses of the rest of the questions from the Medication Inventory; the medication name, strength, units, formulation code, whether or not the container was seen, and other notes. This data set has <u>1 record per medication</u>; therefore, multiple records per Participant. It includes the person's ID to link this dataset to the others. This dataset has no derived variables.

## Medical History (MEDHX) Dataset

This dataset contains information about the Medical History of the Participants, including all diseases the person has/had. This dataset has no derived variables.

## NEO Five-Factor Inventory (NEO) Dataset

This dataset contains the data regarding a personality inventory that examines a person's Big Five personality traits (openness to experience, conscientiousness, extraversion, agreeableness, and neuroticism). This is the full version of the NEO as compared to the short version of the cesdneo data set. This is administered at visit 1 follow up, visit 2 new participants, and anyone who has missing visit 1 NEO data (visitcode is 2, 3, or 4). Derived variables OPENNESS, CONSCIENTIOUSNESS, EXTRAVERSION, AGREEABLENESS and NEUROTICISM are sums of different questions on the form. The SIZE\_OPENNESS, SIZE\_CONSCIENTIOUSNESS, SIZE\_EXTRAVERSION, SIZE\_AGREEABLENESS and SIZE\_NEUROTICISM are the number of non-missing responses for OPENNESS, CONSCIENTIOUSNESS, EXTRAVERSION, AGREEABLENESS and NEUROTICISM,

respectively, which are used to calculate the scores.

#### OPENNESS, CONSCIENTIOUSNESS, EXTRAVERSION, AGREEABLENESS NEUROTICISM, SIZE\_OPENNESS, SIZE\_CONSCIENTIOUSNESS, SIZE\_EXTRAVERSION, SIZE\_AGREEABLENESS and SIZE\_NEUROTOCOSM

\*Run this sas code to generate formatted data and the output file neoall\_formatted.csv is used in the R script below; proc format; VALUE LIKERT 1='Strongly Disagree'

```
2='Disagree'
3='Neutral'
4='Agree'
5='Strongly Agree';
run;
data neo;
set master.neoall;
format worrier -- excellence LIKERT.;;
run;
```

```
proc export data=neo outfile="neoall_formatted.csv" dbms=csv replace; run;
```

#### #R scripts from Boston field site

```
neo.orig <- read.csv(("neoall formatted.csv"), header=T, na.strings=c("", "D","R",</pre>
"N", "U", NA))
#----- Re-level Scales: ------
#Strongly Agree -> 0, Agree -> 1, Neutral -> 2, Disagree -> 3, Strongly Disagree -> 4
#levels(neo.orig$worrier)
#"Agree" "Disagree" "Neutral" "Strongly Agree" "Strongly Disagree"
   levels(neo.orig$worrier)[1] <- 1</pre>
   levels(neo.orig$worrier)[2] <- 3</pre>
   levels(neo.orig$worrier)[3] <- 2</pre>
   levels(neo.orig$worrier)[4] <- 0</pre>
   levels(neo.orig$worrier)[5] <- 4</pre>
#levels(neo.orig$daydream)
   levels(neo.orig$daydream)[1] <- 1</pre>
   levels(neo.orig$daydream)[2] <- 3</pre>
   levels(neo.orig$daydream)[3] <- 2</pre>
   levels(neo.orig$daydream)[4] <- 0</pre>
   levels(neo.orig$daydream)[5] <- 4</pre>
#!!PLEASE NOTE!!: We list code here to get new scales for only two variables: worrier
and daydream. THE SAME CODE TO RE-LEVEL THE SCALE OF THE FOLLOWING 25 VARIABLES
SHOULD BE INCLUDED. We skip those repeated and lengthy codes and do not list them
here.
#25 variables are: stick2, argue, lightheart, selfish, methodical, blue #controversy,
poetry, cynical, alone, takeadv, wastetime, anxious, moods, #moral, cold, optimist,
hardhead, reliable, sad, universe, dislike, #organized, leader, manipulate
#----- REVERSE Scales ------
#Strongly Agree -> 4, Agree -> 3, Neutral -> 2, Disagree -> 1, Strongly Disagree -> 0
   #levels(neo.orig$ppl around)
   #"Agree"
              "Disagree" "Neutral" "Strongly Agree" "Strongly Disagree"
   levels(neo.orig$ppl around)[1] <- 3</pre>
   levels(neo.orig$ppl around)[2] <- 1</pre>
   levels(neo.orig$ppl_around)[3] <- 2</pre>
   levels(neo.orig$ppl around)[4] <- 4</pre>
```

```
levels(neo.orig$ppl around)[5] <- 0</pre>
   #levels(neo.orig$courteous)
   levels(neo.orig$courteous)[1] <- 3</pre>
   levels(neo.orig$courteous)[2] <- 1</pre>
   levels(neo.orig$courteous)[3] <- 2</pre>
   levels(neo.orig$courteous)[4] <- 4</pre>
   levels(neo.orig$courteous)[5] <- 0</pre>
#!!PLEASE NOTE!! We list code here to get new scales for only two variables:
ppl around and courteous. THE SAME CODE TO RE-LEVEL THE SCALE OF THE FOLLOWING 31
VARIABLES SHOULD BE INCLUDED. We skip those repeated and lengthy codes and do not
list them here.
#neat, inferior, laugh, pacing, stress, patterns, talking, cooperate, tense,
#conscientious, action, cleargoals, worthless, newfoods, energy, liked,
#acmplshgoals, angry, cheerful, commitment, discouraged, excitement, fast,
#considerate, productive, helpless, active, curious, ashamed, abstract, excellence
Neuroticism.data <- data.frame( as.numeric(as.character(neo.orig$tense)),
as.numeric(as.character(neo.orig$worthless)),
as.numeric(as.character(neo.orig$anxious)), as.numeric(as.character(neo.orig$angry)),
as.numeric(as.character(neo.orig$discouraged)),
as.numeric(as.character(neo.orig$sad)),
as.numeric(as.character(neo.orig$worrier)),
as.numeric(as.character(neo.orig$inferior)),
as.numeric(as.character(neo.orig$stress)),
as.numeric(as.character(neo.orig$helpless)),
as.numeric(as.character(neo.orig$ashamed)), as.numeric(as.character(neo.orig$blue)))
    names(Neuroticism.data) <- c("tense","worthless","anxious", "angry",</pre>
"discouraged", "sad", "worrier", "inferior", "stress", helpless", "ashamed", "blue")
   Extraversion.data <- data.frame(as.numeric(as.character(neo.orig$lightheart)),</pre>
as.numeric(as.character(neo.orig$talking)),
as.numeric(as.character(neo.orig$leader)), as.numeric(as.character(neo.orig$action)),
as.numeric(as.character(neo.orig$alone)), as.numeric(as.character(neo.orig$energy)),
as.numeric(as.character(neo.orig$cheerful)),
as.numeric(as.character(neo.orig$optimist)),
as.numeric(as.character(neo.orig$fast)),
as.numeric(as.character(neo.orig$ppl around)),
as.numeric(as.character(neo.orig$laugh)), as.numeric(as.character(neo.orig$active)))
     names(Extraversion.data) <-</pre>
c("lightheart","talking","leader","action","alone","energy","cheerful",
"optimist", "fast", "ppl around", "laugh", "active")
   Openness.data <- data.frame(as.numeric(as.character(neo.orig$daydream)),</pre>
as.numeric(as.character(neo.orig$stick2)),
as.numeric(as.character(neo.orig$universe)),
as.numeric(as.character(neo.orig$curious)),
as.numeric(as.character(neo.orig$patterns)),
as.numeric(as.character(neo.orig$controversy)),
as.numeric(as.character(neo.orig$abstract)),
as.numeric(as.character(neo.orig$poetry)),
```

```
as.numeric(as.character(neo.orig$newfoods)),
as.numeric(as.character(neo.orig$moods)),
as.numeric(as.character(neo.orig$moral)),
as.numeric(as.character(neo.orig$excitement)))
     names(Openness.data) <-</pre>
c("daydream","stick2","universe","curious","patterns","controversy","abstract","poetr
y", "newfoods", "moods", "moral", "excitement")
   Agreeableness.data <- data.frame(as.numeric(as.character(neo.orig$cold)),</pre>
as.numeric(as.character(neo.orig$hardhead)),
as.numeric(as.character(neo.orig$courteous)),
as.numeric(as.character(neo.orig$argue)),
as.numeric(as.character(neo.orig$considerate)),
as.numeric(as.character(neo.orig$dislike)),
as.numeric(as.character(neo.orig$selfish)),
as.numeric(as.character(neo.orig$cooperate)),
as.numeric(as.character(neo.orig$manipulate)),
as.numeric(as.character(neo.orig$cynical)),
as.numeric(as.character(neo.orig$takeadv)), as.numeric(as.character(neo.orig$liked)))
     names(Agreeableness.data) <-</pre>
c("cold","hardhead","courteous","argue","considerate","dislike","selfish",
"cooperate", "manipulate","cynical","takeadv","liked")
 Conscientiousness.data <- data.frame(as.numeric(as.character(neo.orig$wastetime)),</pre>
as.numeric(as.character(neo.orig$acmplshgoals)),
as.numeric(as.character(neo.orig$commitment)),
as.numeric(as.character(neo.orig$reliable)),
as.numeric(as.character(neo.orig$neat)), as.numeric(as.character(neo.orig$pacing)),
as.numeric(as.character(neo.orig$productive)),
as.numeric(as.character(neo.orig$organized)),
as.numeric(as.character(neo.orig$methodical)),
as.numeric(as.character(neo.orig$conscientious)),
as.numeric(as.character(neo.orig$excellence)),
as.numeric(as.character(neo.orig$cleargoals)))
    names(Conscientiousness.data) <-</pre>
c("wastetime","acmplshgoals","commitment","reliable"," productive","pacing",
"neat", "organized", "methodical", "conscientious", "excellence", "cleargoals")
   Neuroticism <- apply(Neuroticism.data,1,sum,na.rm=T)</pre>
   Extraversion <- apply(Extraversion.data,1,sum,na.rm=T)</pre>
   Openness <- apply(Openness.data,1,sum,na.rm=T)</pre>
   Agreeableness <- apply(Agreeableness.data,1,sum,na.rm=T)</pre>
   Conscientiousness <- apply(Conscientiousness.data,1,sum,na.rm=T)</pre>
   size.Neuroticism <- apply( is.na(Neuroticism.data)==F,1,sum)</pre>
   size.Extraversion <- apply( is.na(Extraversion.data)==F,1,sum)</pre>
   size.Openness <- apply( is.na(Openness.data)==F,1,sum)</pre>
   size.Agreeableness <- apply( is.na(Agreeableness.data)==F,1,sum)</pre>
   size.Conscientiousness <- apply( is.na(Conscientiousness.data)==F,1,sum)</pre>
  neo.orig <- data.frame(neo.orig, Neuroticism, size.Neuroticism, Extraversion,</pre>
size.Extraversion, Openness, size.Openness,
Agreeableness, size.Agreeableness, Conscientiousness, size.Conscientiousness)
```

## Personal History (PERSHX) Dataset

These variables cover the smoking and alcohol intake histories of the Participants.

#### \_SMOKENOW, \_PIPENOW, and \_PACKYRS

\_SMOKENOW is current smoker. \_PIPENOW is current pipe user. \_PACKYRS is the number of packs smoked per day over the number of years smoked. (# Packs/day \* yrs smoke(d)).

```
length _Smokenow _Pipenow 3;
 merge age(in=a) smoke(in=b);
 by id; if b=1;
 if smoke100=0 or smokequitage>0 or smokequityr>0 then Smokenow=0;
   else Smokenow=smokenow;
 label Smokenow="Current Smoker?";
 if pipe=0 or pipequitage>0 or pipequityr>0 then Pipenow=0;
   else Pipenow=pipenow;
 label _Smokenow="Current Smoker?" _Pipenow="Current Pipe User?";
 format Smokenow Pipenow YND12.;
 if smokenow not in (0,1) then _PACKYRS=.;
 if cigday=. then PACKYRS=.;
 if age =. then _PACKYRS=.;
 if smokenow=1
   then _PACKYRS=(_age - smoke1stage)*(cigday/20);
   else if (smokenow=0 and smoke1styr ne . and smokequityr ne . and cigday ne .)
      then PACKYRS=(smokequityr-smoke1styr)*(cigday/20);
   else if (smokenow=0 and smoke1stage ne . and smokequitage ne . and cigday ne .)
      then _PACKYRS=(smokequitage-smoke1stage)*(cigday/20);
  PACKYRS=round( PACKYRS,.01);
 label _PACKYRS="# Packs/day * yrs smoke(d)";
```

#### \_SUMPACKYRS

\_SUMPACKYRS is similar to \_PACKYRS, above; however, non-smokers are now coded as 0 instead of missing, and the number of years smoked is calculated from the start and quit dates or ages smoked.

\_datayr=year(date);

```
if ((_smokenow=0) and (smoke100 ne 1)) then _sumpackyrs=0; /* never smoker */
if ((_smokenow=1) and (smoke1stage^=.)) then _sumpackyrs=(_age -
smoke1stage)*(cigday/20); /* current smoker, using age */
else if ((_smokenow=1) and (smoke1styr^=.)) then _sumpackyrs=(_datayr -
```

smoke1styr)\*(cigday/20); /\* current smoker, using year \*/

```
if ((_smokenow=0) and (smokenow=0) and (smoke100=1) and (smoke1stage^=.) and (smokequitage^=.) and (cigday^=.))
```

then \_sumpackyrs=(smokequitage - smoke1stage)\*(cigday/20); /\* former smoker, using age start and quit \*/

else if ((\_smokenow=0) and (smokenow=0) and (smoke100=1) and (smoke1styr^=.) and (smokequityr^=.) and (cigday^=.))

then \_sumpackyrs=(smokequityr - smoke1styr)\*(cigday/20); /\* former smoker, using year start and quit \*/

\_yrstart=dobyr+smoke1stage; /\* create year start smoking variable to use for missing former smokers \*/

\_yrend=dobyr+smokequitage; /\* create year end smoking variable to use for missing former smokers \*/

if ((\_smokenow=0) and (smokenow=0) and (smoke100=1) and (\_yrstart^=.) and (smokequityr^=.) and (cigday^=.))

then \_sumpackyrs=(smokequityr - \_yrstart)\*(cigday/20); /\* former smoker, using year start and quit with extrapolated year start\*/

else if ((\_smokenow=0) and (smokenow=0) and (smoke100=1) and (smoke1styr^=.) and (\_yrend^=.) and (cigday^=.))

then \_sumpackyrs=(\_yrend - smoke1styr)\*(cigday/20); /\* former smoker, using year start and quit with extrapolated year end\*/

LABEL \_sumpackyrs="Derived # packs/day \* years smoke(d), for current smokers, former smokers, and non-smokers";

if \_sumpackyrs=-0.2 then \_sumpackyrs=.;

if \_sumpackyrs=-0.6 then \_sumpackyrs=.;

#### \_SMOKE\_CIG, \_SMOKE\_PIPE, and \_SMOKE\_CAT

\_SMOKE\_CIG is current, former, or never cigarette smoker. \_SMOKE\_PIPE is current, former, or never pipe smoker. \_SMOKE\_CAT is current, former, or never cigarette or pipe smoker.

Proc format; Value smkcat 1 = "Never smoked" 2 = "Former smoker" 3 = "Current smoker"; Run;

#### \*\*Cigarette smoking only\*\*;

```
if smoke100=0 then _smoke_cig=1;
else if smoke100=1 and _smokenow=0 then _smoke_cig=2;
else if smoke100=1 and _smokenow=1 then _smoke_cig=3;
**Pipe smoking only**;
if pipe=0 then _smoke_pipe=1;
else if pipe=1 and _pipenow=0 then _smoke_pipe=2;
else if pipe=1 and _pipenow=1 then _smoke_pipe=3;
```

#### **\*\*Cigarette + Pipe smoking\*\*;**

```
if _smoke_cig=1 and _smoke_pipe=1 then _smoke_cat=1;
else if _smoke_cig=1 and _smoke_pipe=2 then _smoke_cat=2;
else if _smoke_cig=1 and _smoke_pipe=3 then _smoke_cat=3;
else if _smoke_cig=2 and _smoke_pipe=1 then _smoke_cat=2;
else if _smoke_cig=2 and _smoke_pipe=2 then _smoke_cat=2;
else if _smoke_cig=2 and _smoke_pipe=3 then _smoke_cat=3;
else if _smoke_cig=3 and _smoke_pipe=1 then _smoke_cat=3;
else if _smoke_cig=3 and _smoke_pipe=2 then _smoke_cat=3;
else if _smoke_cig=3 and _smoke_pipe=2 then _smoke_cat=3;
else if _smoke_cig=3 and _smoke_pipe=3 then _smoke_cat=3;
```

## Physical Function and Activity (PHYSICAL) Dataset

This dataset covers the Physical Exercise form. Variables in the dataset include, activity level, duration, and frequency of exercise. In visit 2, the Pittsburgh Fatigability Scale Test and the Framingham Activity Scale were added to this form. This dataset has no derived variables.

## Performance Measures (PM) Dataset

This dataset includes the results of the Short Physical Performance Battery (SPPB), and the Grip Strength Test.

#### \_TOTSCORE

This derived variable provides one score for the entire SPPB.

WALKSCORE: gives ratings for various values of the walking test.

```
if length eq 1 then do;
    if 0 < shorter < 4.82 then walkscore = 4;
    else if 4.82 <= shorter <= 6.20 then walkscore = 3;
    else if 6.21 <= shorter <= 8.70 then walkscore = 2;
    else if 8.70 < shorter < 60.00 then walkscore = 1;
end;
if length eq 2 then do;
    if 0 < shorter < 3.62 then _walkscore = 4;
    else if 3.62 <= shorter <= 4.65 then walkscore = 3;
    else if 4.66 <= shorter <= 6.53 then walkscore = 2;
    else if 6.53 < shorter < 45.00 then walkscore = 1;
end;
```

totscore = sum (sidescore, semiscore, tdmscore, walkscore, chairscore);

## Prevalence of Disease (PREVDISEASE) Dataset

This dataset was created to derive 11 variables that combine information from the Medical History, Blood Pressure, Blood, and Coded Medications data sets. The 7 derived prevalence disease variables (\_htdis, \_strk, \_lungdis, \_htn, \_diabetes, \_pad and \_cancer) from Visit 1 were completed at Pittsburgh site. DMCC recreated 5 of them (\_htdis, \_strk, \_lungdis, \_pad, and \_cancer) for Visit 2 based on the code used in Visit 1. \_pad was recreated for only Visit 2 new participants because ankle-arm blood pressure ratio (aabprl and aabprr) were not measured for the returning participants. The other two derived variables are recreated when the codedmeds\_atc dataset for Visit 2 is ready, and they are renamed to \_htn\_atc and \_diabetes\_atc. Two more variables for diabetes were derived according to American Diabetes Association (ADA) that used HbA1C as the gold standard for classification of diabetes. They are \_adat2d and \_adat2d\_age\_reported\_detected.

## \_HTDIS, \_STRK, \_LUNGDIS, \_HTN, \_DIABETES, \_PAD, \_CANCER:

proc sort data=clinic.medhx out=medhx; by id; run; proc sort data=clinic.bphr out=bphr; by id; run; proc sort data=blood.blood out=blood; by id; run; proc sort data=codemeds.codedmeds (keep=id htnrx lipidrx dmrx) out=cmeds; by id; run; /\* For visit 2: proc sort data=codemeds.codedmeds atc (keep=id htn diab rename=(htn= htnrx diab=dmrx)) out=cmeds; by id: run: \*/ proc sort data=clinic.sdi (keep=id sex) out=sdi; by id; run; data medhxvars(keep= id \_htdis \_strk \_lungdis); \*data medhxvars(keep= id midx cabg htdis stroke tia strk asth bronch copd lungdis); set medhx: \*create prevalent disease heart disease\*; if (midx=1 or cabg=1) then htdis=1; else htdis=0; \*create prevalent disease stroke\*; if (stroke=1 or tia=1) then \_strk=1; else \_strk=0; \*create prevalent disease lung diseaase\*\*\*\*\*\*; if (asth=1 or bronch=1 or copd=1) then lungdis=1; else lungdis=0; run: proc sort; by id; run; data htn(keep=id htdx); set medhx; run; proc sort; by id; run; data htn1 (keep=id htdx htnrx); merge htn(in=a) cmeds(in=b); by id; if a; run:

```
proc sort; by id; run;
data bp(keep=id htn);
*data bp(keep=id htdx htnrx sys1-sys3 avgsys dia1-dia3 avgdia _htn);
 merge htn1(in=a) bphr(in=b);
 by id; if a;
 avgsys=mean(sys1,sys2,sys3);
 avgdia=mean(dia1,dia2,dia3);
 if ((htdx=1 and htnrx=1)
   or (avgsys >=140)
   or (avgdia \geq 90))
   then htn=1;
   else _htn=0;
run;
proc sort; by id; run;
data diab(keep=id diab); set medhx; run;
proc sort; by id; run;
data diab1(keep=id _diabetes);
*data diab1(keep=id diab dmrx glur diabetes);
 merge diab(in=a) cmeds(in=b) blood(in=c);
 by id; if a;
 if (diab=1 and dmrx=1 or glur >=126)
   then _diabetes=1;
   else _diabetes=0;
run;
proc sort; by id; run;
data pad(keep=id _pad);
*data pad(keep=id aai aabprl aabprr pad);
 set bphr;
 aai = min(aabprl,aabprr);
 if (aai \leq 0.9 and aai > 0) then pad = 1;
 else if aai > 0.9 then _pad=0;
 run:
proc sort; by id; run;
data cancer(keep=id _cancer);
*data cancer(keep=id sex breast leuk colon lung melan skin esophgl pancr ocancer prost _cancer);
 merge medhx(in=a) sdi(in=b);
 by id; if a;
 if (breast=1 or leuk=1 or colon=1 or lung=1 or melan=1 or skin=1 or esophgl=1
  or ocancer=1 or prost=1) and sex=1 then cancer=1;
 if (breast=1 or leuk=1 or colon=1 or lung=1 or melan=1 or skin=1 or esophgl=1
  or ocancer=1) and sex=2 then _cancer=1;
 if _cancer ne 1 then _cancer=0;
 run;
proc sort; by id; run;
data codemeds.prevdisease:
 merge medhxvars(in=a) bp(in=b) diab1(in=c) pad(in=d) cancer(in=e);
 by id; if a;
 if id=" then delete;
```

if \_htdis=. then \_htdis=0; if \_strk=. then \_strk=0; if \_htn=. then \_htn=0; if \_diabetes=. then \_diabetes=0; if \_cancer=. then \_cancer=0; if \_lungdis=. then \_lungdis=0; if \_pad=. then \_pad=0; \*\* At visit 2, aabprl aabprr were measured for new participants, not for returning participants; \*\* Leave \_pad missing for returning participants; run;

#### \_HTN\_ATC, and \_DIABETES\_ATC

```
** codemeds for visit 2 (based on ATC coding scheme);
proc sort data=codemeds atc
out=cmeds(keep=subject htn lipid diab rename=(htn=htnrx lipid=lipidrx diab=dmrx));
by subject; where visitcode in (3,4);
run;
proc sort data=medhxall out=htn(keep=subject htdx); by subject; where visitcode in (3,4); run;
data htn1 (keep=subject htdx htnrx);
merge htn(in=a) cmeds(in=b);
by subject; if a;
run:
proc sort; by subject; run;
proc sort data=bphrall out=bphr(keep=subject sys1 sys2 sys3 dia1 dia2 dia3); by subject; where
visitcode in (3,4); run;
data bp(keep=subject htn atc);
 merge htn1(in=a) bphr(in=b);
 by subject; if a;
 avgsys=mean(sys1,sys2,sys3);
 avgdia=mean(dia1,dia2,dia3);
 if ((htdx=1 and htnrx=1)
   or (avgsys >=140)
```

```
or (avgdia >=90))
then _htn_atc=1;
else _htn_atc=0;
label _htn_atc = `_htn, ATC coding';
run;
```

proc sort data=medhxall out=diab(keep=subject diab); by subject; where visitcode in (3,4); run; proc sort data=bloodall out=blood nodupkey; by subject; where visitcode in (3,4); run;

data diab1(keep=subject \_diabetes\_atc); merge diab(in=a) cmeds(in=b) blood(in=c); by subject; if a; if (diab=1 and dmrx=1 or \_glur\_new >=126)

```
then _diabetes_atc=1;
else _diabetes_atc=0;
label _diabetes_atc = '_diabetes, ATC code';
run;
```

#### \_ADAT2D and \_ADAT2D\_AGE\_REPORTED\_DETECTED

```
proc sort data= medhxall out=medhx(keep=subject visitcode date diab diabage
                                                                               diabnow
rename=(date=medhx_date)) nodupkey;
by subject visitcode;
run;
proc sort data= bloodall out=blood(keep=subject visitcode _glur_new glyhb) dupout=dupblood
nodupkey;
by subject visitcode;
run;
****The data set WORK.DUP has 126 observations (blood re-draw at visit 2);
**visit 2: fasting lipid=1 that indicates blood sample was used to run lipid biomarker test;
data venipv1 venipv2;
set venipall (keep=subject visitcode fast fasting lipid date rename=(date=venip date));
if visitcode=1 and _fast=1 then output venipv1;
if visitcode in (3,4) and fasting_lipid=1 then output venipv2;
run:
data venip; set venipv1 venipv2; run;
proc sort data=venip dupout=dupvenip nodupkey; by subject visitcode; run;
** ATC medication coding scheme;
proc sort data=codedmeds_atc (keep=subject visitcode diab)
     out=cmeds(rename=(diab=dmrx));
by subject visitcode;
run:
proc sort data=sdiall out=sdi(keep=subject visitcode _AGE_REVISED) nodupkey;
by subject visitcode;
run;
data tmp;
merge medhx(in=a) cmeds(in=b) blood(in=c) venip sdi;
by subject visitcode;
if (a or b or c) and visitcode in (1,3,4);
if (_fast>=1 and _glur_new>=126) or glyhb >=6.5 or diabnow=1 or diab=1 or dmrx=1 then _adat2d=1;
  /*if all data in datasets medhx, biomarkers, venipuncture and codedmeds
used for deriving _adat2d is missing, set _adat2d missing */
```

```
else if medhx date in (...R.U.N) and dmrx=. and glyhb=. and glur new=. and venip date in
(.,.R,.U,.N) then _adat2d=.;
else _adat2d=0;
label adat2d='Diabetes? (ADA classification)';
run:
*********(2) derive ADAT2D_AGE_REPORTED_DETECTED;
data v1 v2;
set tmp(keep=subject visitcode _adat2d diabage _age_revised);
if visitcode=1 then output v1;
                                /*visit 1*/
if visitcode in (3,4) then output v2; /*visit 2*/
run;
proc sort data=v1 nodupkey; by subject; run;
proc sort data=v2 nodupkey; by subject; run;
data v1v2;
merge v1(in=in1) v2(in=in2 rename=(visitcode=visitcodev2 _adat2d=_adat2dv2 diabage=diabagev2
_age_revised=_age_revisedv2));
by subject;
if in1 or in2;
/*diabetes: YES for both visit 1 and visit 2*/
/*if more than one age onset was reported, in general, the first age of onset is used, as it is likely closest
to the event.*/
if _adat2d=1 and _adat2dv2=1 then do;
 if diabage>0 then do:
  _ADAT2D_AGE_REPORTED_DETECTED1 = diabage;
  _ADAT2D_AGE_REPORTED_DETECTED2 = diabage;
 end:
 else if diabage\leq 0 and diabage2 > 0 then do;
  if diabagev2 < _age_revised then do;
   _ADAT2D_AGE_REPORTED_DETECTED1 = diabagev2;
   _ADAT2D_AGE_REPORTED_DETECTED2 = diabagev2;
  end:
  if diabagev2 \ge age revised then do;
   ADAT2D AGE REPORTED DETECTED1 = age revised;
   ADAT2D AGE REPORTED DETECTED2 = age revised;
  end;
 end;
 else if diabage\leq=0 and diabagev2\leq=0 then do:
  _ADAT2D_AGE_REPORTED_DETECTED1 = _age_revised; /*if reported age onset missing, use
age at visit 1 in person as age onset*/
  _ADAT2D_AGE_REPORTED_DETECTED2 = age revised:
 end:
end;
/*diabetes: YES for visit 2, not yes for visit 1 */
else if _adat2d ^{=} 1 and _adat2dv2 = 1 then do;
if diabagev2 ^=. then ADAT2D AGE REPORTED DETECTED2 = diabagev2;
```

```
if diabagev2 =. then ADAT2D AGE REPORTED DETECTED2 = age revisedv2; /*if reported age
onset missing, use age at visit 2 in person as age onset*/
end:
/*diabetes: YES for visit 1. NO for visit 2*/
/*According to Ping An, once disbetes was detected, then participant has diabetes*/
else if _adat2d = 1 and _adat2dv2 = 0 then do;
 adat2dv2=1;
 if diabage ^=. then do;
  ADAT2D AGE REPORTED DETECTED1 = diabage;
  ADAT2D AGE REPORTED DETECTED2 = diabage;
 end;
 if diabage = . and _age_revised >0 then do;
  _ADAT2D_AGE_REPORTED_DETECTED1 = age revised;
  ADAT2D AGE REPORTED DETECTED2 = age revised;
 end;
end;
else if adat2d = 1 and adat2dv2 = . then do;
if diabage ^=. then ADAT2D AGE REPORTED DETECTED1 = diabage;
if diabage = . then ADAT2D AGE REPORTED DETECTED1 = age revised;
end;
run;
data tmp2;
set v1v2(keep=subject visitcode adat2d ADAT2D AGE REPORTED DETECTED1
where=(visitcode=1)
rename=( ADAT2D AGE REPORTED DETECTED1= ADAT2D AGE REPORTED DETECTED))
  v1v2(keep=subject visitcodev2 _adat2dv2 _ADAT2D_AGE_REPORTED_DETECTED2
where=(visitcode in (3,4) and adat2d>=0)
    rename=(visitcodev2=visitcode _adat2dv2=_adat2d
ADAT2D AGE REPORTED DETECTED2= ADAT2D AGE REPORTED DETECTED);
label _ADAT2D_AGE_REPORTED_DETECTED = 'Age onset of diabetes: self-reported or detected';
run;
proc sort data=tmp2; by subject visitcode; run;
proc sort data=prevdiseaseall; by subject visitcode; run;
data prevdiseaseall;
merge prevdiseaseall tmp2;
by subject visitcode;
run;
```

## Socio-Demographic Information (SDI) Dataset

This dataset contains the information collected on the Socio-Demographic form.

#### \_AGE

\_AGE is the age when the cognitive assessment form was filled out.

#### \_AGE\_REVISED

\_AGE\_REVISED is the age when the consent form was filled out.

```
_AGE=floor((date-dob)/365.25); **where date is from cogassessall dataset;
_AGE_REVISED=floor((date-dob)/365.25); **where date is from ctifall
dataset;
```

## Spirometry Safety Questionnaire (SPIRO) Dataset

The Spirometry Safety Questionnaire asks background questions that would preclude taking the pulmonary function test, such as major surgery, heart attack, or stroke in the past three months. There are no derived variables in this dataset.

## Spirometry Safety Questionnaire (SPIROMEDS) Dataset

This provides the medication data from question 9b of the Spirometry Safety Questionnaire. This data set has <u>1 record per medication</u>; therefore, multiple records per Participant. It includes the person's ID to link this dataset to the others. This dataset has no derived variables.

## Survival Indices (SURVL\_INDICES) Dataset

This dataset contains derived variables that are indicators of survival and healthy aging.

## **Healthy Aging Index**

Variables related to the healthy aging index (A. Newman, R. Minster, J. Sanders, et al, Pittsburgh) are *HAI*, *HAI\_m*, *HAI\_rg*, *HAI\_m\_rg*, *HAI\_rl*, and *HAI\_m\_rl*.

#### HAI

HAI is the evenly weighted healthy aging index. It is calculated from 5 other variables and thus is only calculated when a LLFS participant has a measurement for all 5 component variables. The 5 component variables are: systolic blood pressure, forced vital capacity, mini-mental state exam, serum creatinine, and serum fasting glucose.

To create the *HAI*, each component variable receives a score of 0 (healthiest tertile), 1 (middle tertile), or 2 (unhealthiest tertile)—with the exception of fasting glucose, for which clinical cutoffs were applied. For systolic blood pressure, if a participant has a physician diagnosis of hypertension or if a participant was using medication for hypertension, they were coded in the unhealthiest tertile (score=2). Similarly for fasting glucose, if a participant has a physician diagnosis of diabetes or if a participant was using medication for diabetes, they were coded in the unhealthiest tertile (score=2). For forced expiratory volume and serum creatinine, separate tertiles were applied to men and women. The scores of the five component variables were then summed for each participant to create the HAI which has a range of 0 (healthiest) to 10 (unhealthiest).

#### HAI Tertile Thresholds

		0	1	2
Systolic blood pressure, m	mHg*	< 126	$\geq$ 126 and <143	≥143
Forced vital capacity, L (women)		≥ 2.61	$< 2.61 \text{ and } \ge 2.14$	< 2.14
	(men)	$\geq$ 3.84	$< 3.84 \text{ and} \ge 3.19$	< 3.19
MMSE, points		> 26	$>$ 23 and $\leq$ 26	≤23
Serum creatinine, mg/dL	(women)	< 0.8	0.8 - 1.0	> 1.0
	(men)	< 1.1	1.1–1.3	> 1.3
Serum fasting glucose, mg	/dL†	< 100	100-125	≥126

\*Physician diagnosis of hypertension or taking anti-hypertensive medication led to score=2 † Physician diagnosis of diabetes or taking medication for diabetes led to score=2

#### HAI\_rg

HAI\_rg is the age, sex, and PC1-10 adjusted residuals of the HAI used for the GWAS.

#### HAI\_m

HAI\_m is the mortality weighted healthy aging index, where each component variable of the HAI is given a mortality optimized weight. Each component score was multiplied by the weight for that component, divided by the sum of all 5 component weights, and multiplied by 5 to obtain the weight for that component. Each component weight was then multiplied to the score for that component and all 5 weighted component scores were summed to calculate the HAI\_m. When we calculated the cox models in CHS for each one point increase in the index we had the betas for mortality. We used the betas as modifiers for the component heritability weights (which before were all equal at 0.20) in the index. That way the weights were optimized for mortality prediction, with the most strongly associated components receiving a greater weight.

HAI m Weights
---------------

	Weight
Systolic blood pressure, mmHg*	0.17085
Forced vital capacity, L	0.38386
MMSE, points	0.42873
Serum creatinine, mg/dL	0.13397
Serum fasting glucose, mg/dL <sup>†</sup>	0.23880

#### HAI\_m\_rg

HAI\_m\_rg is the age, sex, and PC1-10 adjusted residuals of the mortality weighted HAI, HAI\_m, used for the GWAS.

#### HAI\_rl

HAI\_rl is the age and sex adjusted residuals of the HAI used for the linkage analysis.

## HAI\_m\_rl

HAI\_m\_rl is the age and sex adjusted residuals of the mortality weighted HAI, HAI\_m, used for the linkage analysis.

<b>R code (from R. Minster)</b> # Objective: Calculate HAI residuals in LLFS.
# Clean up workspace
rm(list = ls(all = TRUE))
# Set working directory
setwd("/Users/rminster/Documents/Professional/bz-llfs-hai/")
# Load libraries.
# Read in data
$\label{eq:alpha} a <- \mbox{read.csv}("/03-llfs-data/llfsdata_csv_20120413/lungfunc.csv") \\ b <- \mbox{read.csv}("/03-llfs-data/llfsdata_csv_20120413/gtriplet_v2.csv") \\ c <- \mbox{read.csv}("/03-llfs-data/llfsdata_csv_20120413/codedmeds.csv") \\ d <- \mbox{read.csv}("/03-llfs-data/llfsdata_csv_20120413/cogassess.csv") \\ e <- \mbox{read.csv}("/03-llfs-data/llfsdata_csv_20120413/blood.csv") \\ f <- \mbox{read.csv}("/03-llfs-data/llfsdata_csv_20120413/blood.csv") \\ g <- \mbox{read.csv}("/03-llfs-data/llfsdata_csv_20120413/blood.csv") \\ h <- \mbox{read.csv}("/03-llfs-data/llfsdata_csv_20120413/blood.csv") \\ h <- \mbox{read.csv}("/03-llfs-data/llfsdata_csv_20120413/blood.csv") \\ i <- \mbox{read.csv}("/03-llfs-data/llfsdata_csv_20120413/blood.csv") \\ j <- \mbox{read.csv}("/03-llfs-data/llfsdata_csv_20120413/blood.csv") \\ k <- r$
# Merge data into a single file
<pre>data &lt;- merge(a, b, by = "subject") data &lt;- merge(data, c, by = "subject", all.x = TRUE) data &lt;- merge(data, d, by = "subject") data &lt;- merge(data, e, by = "subject") data &lt;- merge(data, f, by = "subject") data &lt;- merge(data, g, by = "subject") data &lt;- merge(data, h, by = "subject") data &lt;- merge(data, i, by = "subject") data &lt;- merge(data, j, by = "subject") data &lt;- merge(data, j, by = "subject") data &lt;- merge(data, j, by = "subject")</pre>
rm(a, b, c, d, e, f, g, h, i, j, k)
# Convert data to correct data type
data\$sys1 <- as.integer(as.character(data\$sys1)) data\$sys2 <- as.integer(as.character(data\$sys2))
data\$creatr <- as.numeric(as.character(data\$creatr))

data\$glur <- as.integer(as.character(data\$glur))  $n \leq nrow(data)$ data sbp <- (data sys1 + data sys2) / 2 data\$fvcl <- data\$fvc / 1000 dataproband <- as.integer(datagen == 2 & data<math>control == 0) data data data data data control == 0) data\$control <- as.integer(data\$gen == 3 & data\$control == 1) data\$gender <- data\$sex.x levels(data\$gender) <- c(NA, "F", "M") # Code from healthiest (0) to least healthy (2) $sbp_t <- rep(NA, n)$ sbp t[datasbp < 126] <- 0  $sbp_t[data$sbp >= 126 \& data$sbp < 143] <- 1$ sbp t[data\$sbp >= 143] <- 2sbp t[datahtnrx == 1] <- 2  $sbp_t[data + 1] < 2$ data <- data.frame(data, sbp\_t)</pre> creat\_t <- rep(NA, n) creat t[datagender = "F" & data creatr < 0.8] <- 0creat\_t[data\$gender == "F" & data\$creatr >= 0.8 & data\$creatr <= 1.0] <- 1  $creat_t[data\$gender == "F" \& data\$creatr > 1.0] <-2$ creat t[datagender == "M" & data creatr < 1.1] <- 0creat t[datagender == "M" & data creatr >= 1.1 & data creatr <= 1.3] <-1 $creat_t[data\$gender == "M" \& data\$creatr > 1.3] <-2$ data <- data.frame(data, creat t)  $fvc_t <- rep(NA, n)$  $fvc_t[data\$gender == "F" \& data\$fvcl >= 2.61] <-0$  $fvc_t[data\$gender == "F" \& data\$fvcl < 2.61 \& data\$fvcl >= 2.14] <-1$  $fvc_t[data\$gender == "F" \& data\$fvcl < 2.14] <-2$ fvc t[datagender == "M" & data fvcl >= 3.84] <-0 $fvc_t[data\$gender == "M" \& data\$fvcl < 3.84 \& data\$fvcl >= 3.19] <-1$  $fvc_t[data\$gender == "M" \& data\$fvcl < 3.19] <-2$ data <- data.frame(data, fvc t)  $gluc_t <- rep(NA, n)$ gluc t[dataglur < 100] <- 0  $gluc_t[data glur >= 100 \& data glur < 126] <- 1$  $gluc_t[data glur >= 126] <-2$ gluc t[data\$X FASTTIME <= 6] <- NA gluc t[datadmrx > 0] <- 2  $gluc_t[data$diab == 1] <- 2$ data <- data.frame(data, gluc t) # From Mike's code -- from comparison of old data, Amy Matteini's codings and CHS  $mmse_t <- rep(NA, n)$  $mmse_t[data\$mmsetot > 26] <-0$ mmse\_t[data\$mmsetot > 23 & data\$mmsetot <= 26] <- 1  $mmse_t[data\$mmsetot \le 23] \le 2$ table(mmse\_t)

```
data <- data.frame(data, mmse_t)</pre>
 # Subset data to what is needed
data <-data[, c("subject", "offspring", "proband", "control", "gen", "fc.x",
"X_AGE", "mmse_t", "sbp_t", "creat_t", "fvc_t", "gluc_t", "gender")]
names(data) <- c("id", "o", "p", "c", "generation", "center",
"age", "mmse", "sbp", "creat", "fvc", "gluc", "sex")
data$center <- as.integer(data$center)</pre>
data center 2 [data center == 2] <- 1
data$center2[is.na(data$center2)] <- 0
data center3 [data center == 3] <- 1
data$center3[is.na(data$center3)] <- 0
datacenter4[data\center == 4] < 1
data$center4[is.na(data$center4)] <- 0
#data[data$c == 1, 3:ncol(data)] <- NA
data <- data[!duplicated(data$id), ]</pre>
 # Equal weighting
w1 <- 0.2
w2 <- 0.2
w3 <- 0.2
w4 <- 0.2
w5 <- 0.2
data hai <- 5 * (w1 * data mmse +
           w2 * data + by +
           w3 * data$creat +
           w4 * datagluc +
           w5 * data$fvc)
 # Mortality-optimized weighting
w1 <- 0.42873
w2 <- 0.17085
w3 <- 0.13397
w4 <- 0.23880
w5 <- 0.38386
w <- sum(w1, w2, w3, w4, w5)
w1 <- w1 / w * 5
w2 <- w2 / w * 5
w3 <- w3 / w * 5
w4 <- w4 / w * 5
w5 <- w5 / w * 5
data$hai_m <- w1 * data$mmse +
         w2 * data + bp +
         w3 * data$creat +
         w4 * data$gluc +
         w5 * data$fvc
table(data$0, data$p, data$c, exclude = NULL, deparse.level = 2)
```

```
names(data)[1] <- "subject"
data <- data[complete.cases(data$hai), ]
m < -lm(hai \sim age + sex, data = data, na.action = na.exclude)
data$hai rl <- residuals(m)
m \le lm(hai m \sim age + sex, data = data,
                        na.action = na.exclude)
data$hai_m_rl <- residuals(m)
write.table(data[data$c == 0, c("subject", "hai_rl", "hai_m_rl")], "3d5c-hai-linkage.csv",
     sep = ",", row = FALSE, quote = FALSE)
pcs <- read.csv("../bl-llfs-genetic-data/2vc-ancestry-pc's/llfseignvec.csv")
i <- read.table("2ve2-genetic-data-ids.txt", header = TRUE)
data <- merge(data[data$subject %in% i$subject, ], pcs[pcs$outlier == 0, ])
m < -lm(hai \sim age + sex + pc1 + pc2 + pc3 + pc4 + pc5 + pc6 + pc7 + pc8 + pc9 + pc
         pc10, data = data, na.action = na.exclude)
m \le tep(m, scope=list(upper = ~ age + sex + pc1 + pc2 + pc3 + pc4 + pc5 + pc6 + pc7 + pc8 + pc9 + pc
                       pc10, lower = ~ age + sex))
summary(m)
data$hai rg <- residuals(m)
m <-lm(hai_m \sim age + sex + pc1 + pc2 + pc3 + pc4 + pc5 + pc6 + pc7 +
                      pc8 + pc9 + pc10, data = data, na.action = na.exclude)
m < step(m, scope=list(upper = ~ age + sex + pc1 + pc2 + pc3 + pc4 + pc5 + pc6 + pc7 + pc8 + pc9 + p
                       pc10, lower = ~ age + sex))
summary(m)
data m rg <- residuals(m)
write.table(data[, c("subject", "hai rg", "hai m rg")], "3d5d-hai-gwas.csv",
      sep = ",", row = FALSE, quote = FALSE)
```

## **Telephone Interview for Cognitive Status (TICS) Dataset**

This data set is part of the Cognitive Assessments. It was used when an in-person visit was not feasible. This dataset has no derived variables.

## Venipuncture (VENIP) Dataset

The Venipuncture dataset contains information about the blood collection, any bleeding disorders, and the shipment information about the tubes. Derived variables were added as well.

## \_FASTTIME, \_FAST, FASTING\_LIPID and FASTING\_CBC

\_FASTTIME is the fasting hours. \_FAST is the fasting status.

#### April 2018 update to venipall data FASTING\_LIPID and FASTING\_CBC

In visit 2, some participants completed more than one venip form. Reasons were that the blood did not process correctly, or at one visit only saliva was collected and then they went back and collected blood for assays. Due to the potential for multiple forms, some participants have more than one row of data in venipall for visit 2, and we needed to create two new variables for these multiple blood draws to assess fasting for the assays. Thus the variables *fasting\_cbc* and *fasting\_lipid* are created to indicate which row of data for visit 2 in the venipall dataset was used for the final values of lipids and cbc and to assess if these values were fasting more than 8 hours or not.

#### How are the *fasting\_cbc* and *fasting\_lipid* created?

Two participants' data from venipall listed in the table below serve as examples. Each participant at visit 2 completed venip form twice, so there are two records for visit 2 in venipall. Reasons for more than one venip form are listed in the right-most column. LLFS Study Design specifies that, with the collected tube #2, i.e. SST tube, the laboratory tests of creatinine, glucose and lipid panel are performed. Also, with the collected tube #3, i.e. EDTA tube, the tests of CBC/diff/platelet and glycosylated hemoglobin are performed. Thus, regardless of value of \_FAST, if tube #2 is not collected, i.e. tube2 = 0, then zero is assigned to fasting\_lipid. Similarly if tube #3 is not collected, zero is assigned to fasting\_cbc.

- (1) Participant 6010 (subject ID): The second venip form for visit 2 was administered on June 6, 2016 for blood samples drawn. Because the fasting hour is 3.33, less than 8 hours, the values of fasting\_cbc and fasting\_lipid are "0". Although the first venip record for visit 2 shows that fasting hour of 16.5, "0" is assigned to fasting\_cbc and fasting\_lipid because there is only saliva was collected for the first venip.
- (2) Participant 22199 (subject ID): Because tube #2 of first draw was not centrifuged, tube #2 was redrawn on March 8, 2016. Laboratory tests of lipid panel were performed on this sample, also the fasting hour is greater than 8 for this redrawn, thus "1" is assigned to fasting\_lipid. As the first tube #2 was not used for tests of lipid panel, therefore fasting\_lipid is "0" in the first venip

form. Laboratory tests of CBC were performed on tube #3 of the first venip, thus "1" is assigned to fasting\_cbc in the first record for visit 2.

subject	visitcode	date	tube1	tube2	tube3	tube4	tube5	tube6	tube7	orag	FASTTIME	_FAST	fasting_lipid	fasting_cbc	Reason for Duplicate Records in Visit 2
6010	1	8-Jan-08	1	1	1	1	1	1		0	14.95	1			
6010	3	25-Apr-16	0	0	0	0	0	0	0	1	16.5	1	0	0	Only Saliva was collected at visit 2 in
6010	3	6-Jun-16	1	1	1	1	1	0	0		3.33	0	0	0	blood draws.
22199	1	5-May-09	1	1	1	1	1	1			10.82	1			
22199	3	15-Feb-16	1	1	1	1	1	1	1		13.42	1	0	1	The SST tube (#2) was not centrifuged:
22199	3	8-Mar-16	0	1	0	0	0	1	1		23	1	1	0	Participant went back for re-draw.

Please note the variables fasting\_cbc and fasting\_lipid are created for visit 2, so for visit 1 the variable \_fast is still used to assess for fasting for visit 1. If you have any questions about how to use this data, please contact the coordinating center.

## What Data Collected per Participant (WHATDATA) Dataset

This data set is an inventory of all the forms and reading center data that were collected for each Participant. This is for visit 1 and visit 2. This dataset has no derived variables

This dataset has no derived variables.

## PHASE II

## Follow Up (FOLLOWUP) Dataset

This dataset contains the information from the Follow Up form. It is completed at every follow up occasion. This dataset has no derived variables.

# **GENOTYPES**

## Annotation (Info and Map) Datasets

The first version of the annotation file was created based on the gene annotation file Illumina provided. The file corresponding to the chip used in genotyping, namely Human Omni 2\_5-8v1 version was used. A large number of markers corresponded with a "kgp" id which was annotating that a particular marker was originated from 1000 Human Genome project. At a later time, an annotation file was provided by CIDR that delivered a correspondence among the "kgp" snps that lacked rs names with now matching rs-names. Therefore, a new annotation is created by merging these new rs names to the existing data. Two file type data are distributed: a) a map file per chromosome; and b) an info file, which have been merged with the NCBI b135 version of dbSNP database to find the most recent version of gene and position annotation. To have a continuation and match with the genotype markers names provided originally by Illumina, the snp names have been kept as the snp identifiers in our database, the improved annotation information by SNP can all be found in the info files for each chromosome.

**NOTE**: The most important connectors in the data are: **markname** in the map, which is a 20 character long identifier for the SNPs and matched with the genotype, genefreq and info files. The corresponding variable with no padding "XXXXXX" is **snpname**. The next important variable is **newmarkname**, which represent a variable with the most rs-names known in this chip. The corresponding variable with no padding "XXXXXX" is **newsnpname**. The rest of the variables are annotations, such as chromosome, position, gene name, role of the SNP. It will be of interest that one uses the information that comes from the latest NCBI dbSNP annotated for example as **b135\_chrom**, **b135\_pos** etc.

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nugoz		19	kap10011626X0000000X	kap10011626	rs5994840X00X000X00X	rs5994840	22	3 kgp	o1000167 kgr	p1000167	rs78613736	rs78613736		22 4	47614430	:	22 476144	30
diffPosNearGene		20	kap10012824XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	kap10012824	rs5759218X0000000000	rs5759218	22	4 kgp	o1000198 kgp	p1000198	rs5995267X	rs5995267		22 3	36633107		22 366331	07
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il_alleles		25	km10023679XXXXXXXXXXXX	kgp10023679	rs5766790XXXXXXXXXXXXXX	rs5766790	22	10 kgr	1000533 kg	1000533	re62225084	m62225064		22 1	18122841		22 233430	41
/ ill_build		26	kap10024252XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	kap10024252	rs78090161XXXXXXXXXXX	rs78090161	22	11 kgr	1000609 kgr	n1000609	rs77004047	rs77004047		22 3	39708067		22 397080	67
III_sourceversion		27	kap10026386XXXXXXXXXXXXXX	kap10026386	rs4823126X00XX00XX00X	rs4823126	22	12 kgr	1000663 kg	1000663	rs9623543X	rs9623543		22	12622369		22 426223	69
II_SourceStrand		28	kgp10027123XXXXXXXXXXXXXXX	kgp10027123	rs74536383XXXXXXXXXXXXX	rs74536383	22	13 kg	1000665 kg	01000665	rs12628783	rs12628783		22 4	10586069		22 405860	69
il name		29	kon10027484XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	kgp10027484	rs28496879XXXXXXXXXXXXXX	rs28496879	22	14 kgp	01000733 kg	01000733	rs78085690.	rs78085690		22 4	19438037		22 494380	37
ilgene hugo		30	kgp10027508XXXXXXXXXXXXX	kop10027508	rs4074029XXXXXXXXXXXXXXXXX	rs4074029	22	15 kgp	01000838 kg	p1000838	rs882754XX	rs882754		22 3	27201909	:	22 272019	09
L illgene_GeneLoc		31	km10029039XXXXXXXXXXXXXX	kgp10029039	rs73427858XXXXXXXXXXXXXX	rs73427858	22	16 kgp	01000978 kgp	p1000978	rs17807076	rs17807076		22	17582776		22 175827	76
illgene_ExonLoc	-	32	kgp1002909XXXXXXXXXXXXXXX	kgp10020000	rs5759527XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	rs5759527	22	17 kgp	01001020 kgp	p1001020	rs80056303	rs80056303		22 3	36848023		22 368480	23
	-	33	kap10020252XXXXXXXXXXXXX	kgp1002000	re130623XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	re130623	22	18 kgp	01001091 kgp	p1001091	rs5748924X	rs5748924		22	17647879		22 176478	79
<ul> <li>Rows</li> </ul>		24	kon10029541XXXXXXXXXXXXXX	kap10020202	rs6004754XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	rs6004754	22	19 kgp	o1001162 kgp	p1001162	rs5994840X	rs5994840		22 3	22080878	1	22 220808	78
All rows	31,048	20	kon10020758XXXXXXXXXX	kap10020341	re16001082XXXXXXXXXXXX	re16001082	22	20 kgp	o1001282 kgp	p1001282	rs5759218X	rs5759218		22 4	43614588		22 436145	88
Selected	0	30	kap10020700/00/00/00/00/00/00/00/00/00/00/00/00	kap10020/00	re4585126YYYYYYYYY	re4585126	22	21 kgp	o1001612 kgr	p1001612	rs10483238	rs10483238		22 4	48081385		22 480813	85
Excluded	0	30	kap10022222222222222222222222222222222222	kap10022	r=50001561VVVVVVV	134000120	24	22 kgp	o1001629 kgp	p1001629	kgp1001629	kgp1001629	94	22 4	43159382			
Labelled	0	37	kap100324067777777777777	kap10032	r=70424601VVVVVVVVVVV	15000/1001	22	Rows 1-1	10000 of 31048	Filter: Off	Sort: none							
CONCINCI			III	- ngp10034007	131043400170000000000	1370434001	- 24	\\mercurv	5.dsa.wustl.edu	ı\dsa500\IIf	s\data\sasne	no final\c22\m	ap22.sas7bda	t				

The above is a picture combination of a view of map22.sas7bdat and info22.sas7bdat. Following are two tables that summarize the work on markers for the maps and annotation files.

No	ilrs	ilkgp	other	total	new_rs	still_kgp	other	total	b135	b131	illonly	total	diff
1	57,135	126,907	30	184,072	168,169	15,873	30	184,072	167,408	109	16,162	183,679	393
2	55,980	138,146		194,126	178,112	16,014		194,126	177,299	118	16,249	193,666	460
3	45,960	117,712		163,672	150,012	13,660		163,672	149,541	97	13,872	163,510	162
4	39,198	113,647	1	152,846	139,835	13,010	1	152,846	139,260	86	13,200	152,546	300
5	40,847	104,603	3	145,453	133,109	12,341	3	145,453	132,748	90	12,547	145,385	68
6	54,996	99,673	17	154,686	142,373	12,296	17	154,686	141,543	102	12,725	154,370	316
7	36,782	92,253	37	129,072	118,479	10,556	37	129,072	117,926	72	10,813	128,811	261
8	35,929	89,586		125,515	115,631	9,884		125,515	115,345	90	10,026	125,461	54
9	31,739	71,268	4	103,011	95,458	7,549	4	103,011	95,078	57	7,676	102,811	200
10	37,889	81,519		119,408	109,694	9,714		119,408	109,364	65	9,901	119,330	78
11	35,123	80,970	2	116,095	106,430	9,663	2	116,095	106,099	91	9,816	116,006	89
12	34,478	78,219	25	112,722	103,177	9,520	25	112,722	102,687	92	9,732	112,511	211
13	27,034	56,447	2	83,483	76,633	6,848	2	83,483	76,327	43	7,077	83,447	36
14	22,510	54,000		76,510	70,360	6,150		76,510	70,166	50	6,253	76,469	41
15	21,078	51,211	5	72,294	66,467	5,822	5	72,294	66,291	39	5,916	72,246	48
16	21,821	54,787	2	76,610	70,874	5,734	2	76,610	70,669	35	5,838	76,542	68
17	19,260	47,125	2	66,387	61,111	5,274	2	66,387	60,915	39	5,387	66,341	46
18	21,117	47,435		68,552	63,465	5,087		68,552	63,325	40	5,163	68,528	24
19	13,853	33,880		47,733	43,767	3,966		47,733	43,557	40	4,088	47,685	48
20	17,802	38,739	1	56,542	52,731	3,810		56,541	52,581	25	3,885	56,491	51
21	9,940	22,135		32,075	29,470	2,605		32,075	29,389	16	2,644	32,049	26
22	9,837	23,473		33,310	31,035	2,275		33,310	30,899	13	2,323	33,235	75
	690,308	1,623,735	131	2,314,174	2,126,392	187,651	130	2,314,173	2,118,417	1,409	191,293	2,311,119	3,055

chrom	LIBNAME	NAME	remobs	NOBS
1	C1	markname	379	176,754
2	C2	markname	444	187,627
3	C3	markname	154	158,475
4	C4	markname	291	148,058
5	C5	markname	66	140,918
6	C6	markname	288	148,707
7	C7	markname	251	123,975
8	C8	markname	50	121,422
9	C9	markname	188	98,900
10	C10	markname	75	114,826
11	C11	markname	85	111,619
12	C12	markname	198	108,540
13	C13	markname	36	80,896
14	C14	markname	39	73,798
15	C15	markname	44	69,671
16	C16	markname	60	72,736
17	C17	markname	46	62,495
18	C18	markname	24	66,377
19	C19	markname	46	43,783
20	C20	markname	46	54,055
21	C21	markname	23	30,798
22	C22	markname	70	31,048
Total			2,903	2,225,478

Final count of markers included in the data follows:

## Info Datasets Variables

**markname**: Illumina provided locus name (rs number, if available) padded with "XXX" to 20 characters long

**snpname**: Illumina provided locus name (rs number, if available)

**newmarkname**: CIDR provided locus name (rs number, if available) padded with "XXX" to 20 characters long

newsnpname: CIDR provided locus name (rs number, if available)

**ill\_chrom**: Illumina provided chromosome number

**ill\_pos**: Illumina provided base pair position

b135\_chrom: dbSNP Build 135 chromosome number

b135\_pos: dbSNP Build 135 base pair position

**b131\_pos**: dbSNP Build 131 base pair position

b131\_chrom: dbSNP Build 131 chromosome number

**rs**: SNP rs number if found

alleles: dbSNP Build 135 alleles strand: dbSNP Build 135 strand hugo: dbSNP Build 135 gene symbol role: dbSNP Build 135 SNP function class

ABBREV	DESCRIPTION
cds-synon	synonymous change. ex. rs248, GAG->GAA, both produce amino
	acid: Glu
intron	intron. ex. rs249.
cds-reference	contig reference
synonymy unkno	owncoding: synonymy unknown
nearGene-3	within 3' 0.5kb to a gene. ex. rs3916027 is at NT_030737.9
	pos7669796, within 500 bp of UTR starts 7669698 for
	NM_000237.2.
nearGene-5	within 5' 2kb to a gene. ex. rs7641128 is at NT_030737.9
	pos7641128, with 2K bp of UTR starts 7641510 for NM_000237.2.
STOP-GAIN	changes to STOP codon. ex. rs328, TCA->TGA, Ser to terminator.
missense	alters codon to make an altered amino acid in protein product. ex.
	rs300, ACT->GCT, Thr->Ala.
STOP-LOSS	changes STOP codon to other non-stop codon
frameshift	indel snp causing frameshift.
cds-indel	indel snp with length of multiple of 3bp, not causing frameshift.
UTR-3	3 prime untranslated region. ex. rs3289.
UTR-5	5 prime untranslated region. ex. rs1800590.
splice-3	3 prime acceptor dinucleotide. The last two bases in the 3 prime end
	of an intron. Most intron ends with AG.ex.rs193227 is in acceptor
	site.
splice-5	5 prime donor dinucleotide. 1st two bases in the 5 prime end of the
	intron. Most intron starts is GU. ex.rs8424 is in donor site.

hugo2: dbSNP Build 135 other strand overlapping gene symbol

role2: dbSNP Build 135 other strand overlapping SNP function class

- **diffPosNearGene**: Distance (bp) to the nearest gene. = 0 if SNP on the gene, < 0, if with lower position (on upstream), > 0 if with higher position (on downstream)
- **Newhugo**: The nearest gene name in () if hugo is missing. If distance to the nearest gene (i.e. diffPosNearGene) > 5 kbp, postfix "\_beyond" to gene name.

IlmnStrand: Illumina provided strand

ill\_alleles: Illumina provided alleles

**ill\_build**: Illumina provided build version

ill\_sourceVersion: Illumina provided source version

ill\_sourceStrand: Illumina provided source strand

**ill\_refStrand**: Illumina provided reference strand

illgene\_hugo: Illumina provided gene symbol

illgene\_GeneLoc: Illumina provided gene location illgene\_ExonLoc: Illumina provided exon location illgene\_CodingStatus: Illumina provided coding status cidr\_chrom: CIDR provided chromosome number cidr\_pos: CIDR provided base pair position P\_HWE: SNP Hardy Weinberg Equilibrium P Value Callrate: SNP genotyping callrate coded\_all: GWAS coded allele noncoded\_all: GWAS other allele coded\_af: Allele frequency of the coded allele

## Anonymous Genotypes (GANON) Datasets

These represent the genotype datasets, organized by chromosome. There is one record per Subject and the columns represent the SNP markers. Each cell contains the genotype coded as allele1/allele2, in numerical representation where 1=A, 2=C, 3=G, and 4=T. These datasets are provided in two different formats, SAS and CSV. In addition, we provide CSV formatted subsets, split for conducting parallel programming.

•]ganon_22	× •	subject	Kgp100001XXXX	kap1000650XXXXXXXXXX	kap10001673XXXXXXXXXXX	kap10001984XXXXXXXXXXXX	kap10004393XXXXXXXXXXXX	xgp1000446XXX	kap10004478XXXXXXXXXXXX	kap10004808XXXXXXXXXXXXX	kap10005
		subject	1/1	4/0	Kgp Tovo Tor o Joso Joso A	0.0	10	4/4	2/2	1/1	ageroood
	1	8	1/1	4/2	1/1	2/2	1/3	1/1	2/2	1/1	3/3
	4	9	1/1	4/2	1/1	2/2	3/3	1/1	212	1/3	1/3
<ul> <li>Columns (31049/0)</li> </ul>	3	10	1/1	4/2	1/1	2/2	1/3	1/1	2/2	1/3	1/3
d subject	4	27	1/3	4/2	1/1	2/2	1/3	1/1	4/2	1/1	3/3
	5	i 46	1/3	4/2	1/1	2/2	1/1	1/1	2/2	1/3	3/3
kap10000650XXXXXXXXXXXX	6	47	1/1	4/2	1/1	2/2	1/3	1/3	2/2	1/3	1/3
kgp10001673XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	7	50	1/3	4/2	1/1	2/2	1/1	1/1	2/2	1/3	3/3
L kgp10001984XXXXXXXXXXX	8	51	1/3	4/2	1/1	2/2	1/3	1/1	2/2	1/1	3/3
L kgp10004393XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	9	61	1/1	4/2	1/1	2/2	1/1	1/1	2/2	1/3	1/3
L kgp1000446XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	10	81	1/3	4/2	1/1	2/2	1/3	1/1	2/2	1/1	1/3
L kgp10004478XXXXXXXXXXXXX	11	85	3/3	2/2	1/1	2/2	3/3	1/1	2/2	1/3	1/1
L kgp10004808XXXXXXXXXXX	12	86	1/3	4/2	1/1	2/2	1/3	1/1	2/2	1/3	1/3
kgp10005335XX000XX000X	13	87	1/1	4/2	1/1	2/2	1/3	1/1	2/2	1/3	3/3
kgp10005418XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	14	88	3/3	4/2	1/1	2/2	1/3	1/1	2/2	1/3	1/1
kgp10006090XXXXXXXXXXXX	15	80	1/1	2/2	1/1	2/2	1/3	1/1	2/2	1/3	1/1
kgp10006633XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	10	00	1/2	4/2	1/1	2/2	1/2	1/1	2/2	1/1	1/2
kgp10006653XXXXXXXXXX	10	01	1/0	4/2	4/4	1/2	1/3	4/4	2/2	1/1	4/4
kgp10007333XXXXXXXXX	17	91	1/3	4/2	1/1	1/2	10	1/1	2/2	1/3	1/1
kap10008382XXXXXXXXXX	18	94	1/3	4/2	1/1	2/2	1/3	1/1	2/2	1/1	3/3
kap10010207XXXXXXXXXXX	19	102	1/3	4/2	1/1	2/2	1/3	1/1	2/2	1/1	1/3
kap10010207XXXXXXXXX	20	104	1/3	4/2	1/1	2/2	1/1	1/1	2/2	1/1	3/3
kap10011626XXXXXXXXXX	21	105	1/3	4/2	1/1	2/2	1/1	1/1	2/2	1/1	1/3
kap10012824XXXXXXXXXXXXX	22	106	1/1	4/2	1/1	2/2	1/3	1/1	2/2	1/1	1/3
kgp1001612XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	23	112	1/1	4/2	1/1	2/2	1/3	1/1	2/2	1/1	1/3
L kgp10016294XXXXXXXXXXXX	24	113	1/1	4/2	1/1	2/2	1/3	0/0	2/2	1/3	1/1
L kgp1001909XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	25	115	1/3	2/2	1/1	2/2	3/3	1/1	2/2	1/1	3/3
L kgp10019876XXXXXXXXXXXX	26	117	1/1	2/2	1/1	2/2	3/3	1/1	2/2	1/3	3/3
L kgp10023679XXXXXXXXXX	27	118	1/3	4/2	1/1	2/2	3/3	1/1	2/2	1/3	1/3
kgp10024252XXXXXXXXXXXXX	28	121	1/3	4/2	1/1	2/2	1/1	1/1	2/2	1/3	3/3
kgp10026386XXXXXXXXXXXXX	29	122	1/3	4/2	1/1	2/2	1/3	1/1	2/2	1/1	3/3
kgp10027123XX00XX00X	30	123	1/1	4/4	1/1	2/2	1/1	1/1	2/2	1/1	1/3
kgp10027484XXXXXXXXXXXX	31	124	1/3	4/2	1/1	2/2	1/3	1/1	2/2	1/1	3/3
Kgp10027508XXXXXXXXX	32	140	1/3	2/2	1/1	2/2	3/3	1/1	2/2	1/1	3/3
kgp10029039XXXXXXXX	32	140	1/2	2/2	4/4	2/2	0/0	4/4	2/2	4/4	4/4
Kgp1002909XXXXXXXXXX	33	442	1/3	2/2	1/1	2/2	4/2	4/4	2/2	1/1	4/2
<ul> <li>Rows</li> </ul>	34	143	10	2/2	1/1	212	0.0	111	212	111	10
All rows 4,693 ^	35	148	1/3	2/2	1/1	2/2	3/3	1/1	2/2	1/1	1/3
Selected 0	36	151	1/1	2/2	1/1	2/2	1/3	1/1	2/2	1/1	1/3
Excluded 0 =	37	161	1/1	2/2	1/1	2/2	1/3	1/1	2/2	1/3	3/3
Hidden 0	38	162	1/1	2/2	1/1	2/2	1/3	1/1	2/2	3/3	3/3 *

## Gene Frequency (GENEFREQ) Datasets

These sets of data (one per chromosome) contain the marker names (MARKNAME), numeric representations of the alleles (ALLELES), the frequency of each allele for each marker in the sample (PERCENT), expressed as a percent, and the number of subjects that carried that allele (COUNT) and total of counts. These data sets have 1 record per allele, usually two per marker. In case a marker is nonpolymorphic, then one will see 1 allele with 100 as its percentage.

genetreqc22 ₽	۹ 🗸 💌						
		markname	allele	COUNT	PERCENT	total	
	1	kgp10069433XXXXXXXXXXX	2	8837	94.191	9382	
	2	kgp10069433XXXXXXXXXXX	4	545	5.808996	9382	
	3	kgp10075858XXXXXXXXXX	1	3861	41.19718	9372	
	4	kgp10075858XXXXXXXXXXX	2	5511	58.80282	9372	
	5	kgp10080081XXXXXXXXXXXX	1	1135	12.15724	9336	
	6	kgp10080081XXXXXXXXXX	3	8201	87.84276	9336	
	7	kgp10085808XXXXXXXXXX	1	1324	14.10612	9386	
	8	kgp10085808XXXXXXXXXX	3	8062	85.89388	9386	
	9	kgp10096682XXXXXXXXXX	1	5990	63.81845	9386	
Columns (5/0)	10	kgp10096682XXXXXXXXXXXXX	3	3396	36.18155	9386	
markname	11	kgp10134312XXXXXXXXXXXX	1	242	2.578858	9384	
	12	kgp10134312XXXXXXXXXX	3	9142	97.42114	9384	
	13	kgp10139604XXXXXXXXXX	2	1715	18.27968	9382	
PERCENT	14	kgp10139604XXXXXXXXXX	4	7667	81.72032	9382	
🚄 total	15	kgp10141318XXXXXXXXXXX	2	2578	28.00956	9204	
	16	kgp10141318XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX	3	6626	71.99044	9204	
	17	kgp10444672XXXXXXXXXXX	2	2642	28.22047	9362	
	18	kgp10444672XXXXXXXXXXX	4	6720	71.77953	9362	
	19	kgp10536361XXXXXXXXXX	1	6376	68.25091	9342	
	20	kgp10536361XXXXXXXXX	3	2966	31.74909	9342	
	21	kgp1057882XXXXXXXXXXXX	2	8416	91.26003	9222	
	22	kgp1057882XXXXXXXXXXXXXXX	4	806	8.73997	9222	
	23	kgp10680932XXXXXXXXXX	2	9261	98.68926	9384	
Rows	24	kgp10680932XXXXXXXXX	4	123	1.310742	9384	
All rows 59,809	25	kgp10729458XXXXXXXXXX	3	9350	100	9350	
Selected 0	26	kgp10898049XXXXXXXXXX	2	4312	45.95055	9384	
Excluded 0	27	kap10898049XXXXXXXXXX	4	5072	54.04945	9384	
Hidden 0	28	kgp10935731XXXXXXXXXXXX	2	666	7.095674	9386	
Labelled 0	29	kap10935731XXXXXXXXXX	4	8720	92 90433	9386	

## GTRIPLET and TRIPLET\_visit2 Dataset

The GTRIPLET dataset reflects the pedigree structures corrected using genetic information and GRR (Graphical Representation of Relationships). Therefore, it is the preferred pedigree structure for analysis. A triplet is the person, his/her mother, and his/her father. This is all the necessary information needed to determine relatedness.

**SUBJECT:** the de-identified, unique identifier for each Participant. It is a 5 digit number. Datasets with one obs/subject are uniquely identified by ID and can be merged/linked using this variable.

**MOMSUBJ:** the SUBJECT number of the person's mother

**DADSUBJ:** the SUBJECT number of the person's father

Proband\_status: the index indicates a subject who is the proband in a pedigree.

**gpedid:** is the preferred indicator of family membership since it is derived using genetic information.

deceased: vital status.

twinrelatn: twin relationship, MZ or DZ.

relative: the Subject is genetically related to the proband.

**control:** the Subject is married into the Proband's offspring generation. This person married an offspring of the Proband.

gen: Generation Number.

- 1 = Proband's Parents Generation
- 2 = Proband's Generation
- 3 = Proband's Offspring Generation
- 4 = Proband's Grandchildren Generation

In the pedigree plot (in the figure on the next page), the color code reflects the values of relative and control. The diagonal line indicates a deceased Subject. The black arrow points to the Proband. Circles are females, squares are males, and diamonds are dummy, placeholder children, to indicate the relationship of a spouse pair without biological children between them.

