

Appendix A

Data Collection Forms

- **TS1:** LLFS Family Longevity Selection Score (FLoSS) Instrument - Proband
- **TS2:** LLFS Proband Relative Contact Information Form
- **TS3:** LLFS Pedigree Information Form (PIF)
- **TS1a:** LLFS Telephone Screener – Family Member
- **TS1a-c:** LLFS Telephone Screener – Control Subjects
- **TS1c:** LLFS Family Longevity Score (FLoSS) Instrument – Control Subjects
 - **TS1c:** LLFS FLoSS Instrument – Control Subjects [**UP/BU Pilot**]
- **Panel 1:** Consent Tracking & Interview Feasibility
 - Generic Consent
 - Boston Consent
 - Columbia Consent
 - Pittsburgh Consent
- **Panel 1a:** Assessment for Capacity to Provide Informed Consent
- **Panel 1b:** Criteria for Proxy-Based Interviews
- **Panel 2:** Socio-Demographic Information
 - US Form
 - Denmark Form
- **Panel 3:** Physical Function and Activity
- **Panel 4:** Personal History Interview
- **Panel 5:** Medical History Interview
- **Panel 6:** Medication Inventory Form
- **Panel 7:** Cognitive Assessments
 - **Panel 7a:** NACC UDS Forms
 - **Panel 7b:** Telephone Interview for Cognitive Status (TICS)
 - **Panel 7c:** Informant-Based Date of Onset Interview
- **Panel 8:** Performance Measures
- **Panel 9:** Blood Pressure, Heart Rate, Height, Weight and Waist Circumference
- **Panel 10:** Spirometry
- **Panel 11:** Mood and Personality Assessment
 - CES-D
 - NEO Five-Factor Inventory
- **Panel 12:** Pedigree Family Structure Worksheet*
- **Panel 13:** Blood Collection: Venipuncture Form
- **Panel 14:** Participant Contact Information Form
- **Panel 15:** LLFS Forms & Testing Checklist Worksheet*
- **Panel 16:** LLFS Follow-Up Telephone Contact
- **Panel 17:** Remote Travel Request Form (Worksheet; *Not to be administered to participants*)*

*Worksheet indicates that form is not to be data-entered.



Appendix B

Response Forms for Questionnaires

Father or Mother

Step-Father or Step-Mother

Husband or Wife

Son or Daughter

Step-Son or Step-Daughter

Brother or Sister

Half-Brother or Half-Sister

Step-Brother or Step-Sister

Uncle or Aunt

Nephew or Niece

Cousin

In-Law

Other (Please Specify)

Yes, Mexican American/Chicano

Yes, Puerto Rican

Yes, Cuban

Yes, Other Spanish/Hispanic Latino

No, Not Spanish/Hispanic/Latino

White

Black or African-American

American Indian or Alaska Native

Asian

Native Hawaiian or Other Pacific Islander

Other (Please Specify)

Never Went To School

No Schooling Completed

Nursery School to 4th Grade

5th Grade or 6th Grade

7th Grade or 8th Grade

9th Grade

10th Grade

11th Grade

12th Grade – No High School Diploma or GED

High School Graduate – Diploma or GED

Vocational Training
(Technical, Agricultural Trade or Craft)

Some college credit, but less than 1 Year

1 or more years of College – No Degree

Associate Degree (AA, AS)

Bachelor's Degree (BA, AB, BS)

Master's Degree
(MA, MS, MENG, MED, MSW, MBA)

Professional Degree
(MD, DDS, DVM, LLB, JD)

Doctorate Degree (PhD, EdD)

LESS THAN \$5,000

\$5,000 to \$10,000

\$10,000 to \$15,000

\$15,000 to \$20,000

\$20,000 to \$35,000

\$35,000 to \$50,000

\$50,000 to \$75,000

\$75,000 to \$100,000

\$100,000 +

Very Easy

Easy

OK

Hard

Very Hard

Less than \$100,000

\$100,000 to \$250,000

\$250,000 to \$500,000

Equal to or more than \$500,000

Daily

4 to 6 Times per Week

2 to 3 Times per Week

Once Per Week

Less than Once Per Week

A Little Difficulty

Some Difficulty

A Lot of Difficulty

I Am Unable to Do It

None, Do Not Drink at All

Occasional Drink, but Less than Once per Week

1-3 Drinks per Week

4-7 Drinks per Week

8-14 Drinks per Week

15-21 Drinks per Week

22-27 Drinks per Week

28 or More Drinks per Week

No Need or Not Necessary

Don't Care for It or Dislike It

Medical or Health Reasons

Religious or Moral Reasons

Recovering Alcoholic

Family Member an Alcoholic or Problem Drinker

Costs too Much

Other (Please Specify)

Rarely or none of the time (less than 1 day)

Some or a little of the time (1 to 2 days)

A moderate amount of time (3 to 4 days)

Most of the time

Strongly disagree

Disagree

Neutral

Agree

Strongly agree

Appendix C

'Sample' Informant Consent

Consent to Participate in a Research Study (Informant)

The purpose of this consent form is to provide you with the information you need to consider in deciding whether to participate in this research study.

Study Title: LONG LIFE Family Study (LLFS)

IRB Study Number: XXXXXXXXXXX

Purpose of Study: You are being asked to participate in a research study on exceptional aging as an informant for _____, (participant). Exceptional longevity (which means living until a very old age) and good health in old age appear to run strongly within some families. The purpose of the LONG LIFE Family Study is to find out why some people are able to live into very old age. By studying long-lived families, we hope to learn how much of longevity is because of genes that run in families and how much is due to other things, such as healthy life habits. Your participation in this research study may help us learn how some people are able to live to a very old age, spending most of their lives in good health and avoiding many age-related diseases.

We expect to enroll approximately 1,000 families (or approximately 5,500 people across the 4 sites participating in the LONG LIFE Family Study. The study sites are Columbia University in NY, the University of Pittsburgh in Pennsylvania, Boston University in Massachusetts and the University of Southern Denmark in Denmark. Washington University, St. Louis will serve as the Coordinating Center for this project. You would be a part of this study that is based at Columbia University. There will be approximately 275 families/1500 people entered into this study at this site. This study is supported by a grant from the National Institute on Aging.

Alternatives to Participation: This is not a treatment study. Information being collected is for research purposes only and is used to learn more about the health of older adults and their families, not about you, and will not provide information that will be medically useful to you. The alternative to participating would be simply not to participate.

Our study research staff will be available to answer questions about the results and to provide referrals to you or the participant if you or s/he does not have your/her/his own physician.

The medical assessments and examinations done as part of this study will not provide you or the participant with the same kind of clinical information you would obtain from your/his/her doctor during a medical visit. We can help you/him/her with a referral if you are seeking medical testing.

Study Procedures: If you decide to take part in this research study as an informant for _____ (participant), you will undergo the following procedures:

In-Person Visit

A home visit examination will take place at a time that is convenient for you and [redacted] (participant). If you prefer a clinic exam, a visit at Columbia University Medical Center will be offered for your convenience. If we are unable to see you in person, some of the interviews and assessments may be conducted over the phone.

During the visit, a trained clinical staff member will obtain medical and personal information about the participant for whom you are the informant. You will be asked to answer questions related to his/her current and past medical history, medication use, daily living activities, physical activity as well as **his/her** health habits (i.e, smoking and alcohol use). You will also be asked demographic questions, such as how many years of education the participant has had, when and where s/he was born, and his/her occupation. You will be asked for a copy of his/her birth certificate or other evidence of his/her birth date. If s/he does not have a copy of his/her birth certificate, you will be asked for information that will help us obtain one. Additionally, you will be asked about **his/her** marital status, current living situation, as well as his/her maximal household income and assets. You may be asked about the ages of his/her children, brothers, sisters, parents where they were born and, if they have died, what they died from and any health conditions they may have had. This information will help us draw his/her family's pedigree (family tree). We may ask you for a representative of his/her immediate family to verify the correctness of this information. Other questionnaires will include a paper and pencil tests which will be performed on the participant to test his/her ability to process and recall information and a personality assessment. You or the participant may be asked to complete some of these questionnaires on your own and send them back to us in a pre-paid envelope. We may call you after we receive your completed questionnaire just to clarify some of your answers if needed. Some of these questionnaires that will be sent to you will include many of the same questions asked of you above, but this time, you will be asked about his/her family members so that you may help us collect information about the background and health of his/her family.

Measurements of the participant's weight, height, waist circumference, knee height, arm span, heart rate and ankle/arm blood pressure will be attempted. S/he will also be asked to blow into a tube to measure his/her lung function and to perform some simple physical tasks such as standing from of a chair, gripping an object to measure hand strength and walking a short distance to assess physical function. Additionally, s/he will be asked to perform a series of movements to test his/her balance. This examination can be completed in approximately 2 hours.

Lastly, you may be asked to provide us with the names of 2 close family members of the participant who we can contact to inquire about his/her status if we are unable to contact you or the participant at a later date for the telephone follow-up visit (mentioned on page 4 of this consent form).

Blood Tests

The participant will be asked to have a blood sample drawn after obtaining his/her and your consent, as the informant. Since this study is hoping to identify genes associated with longevity that may run in families, the blood sample is a very important part of the study. S/he will be asked to go without food for a period of 8 hours prior to collection of a blood sample. For his/her convenience, a separate visit to the home by a phlebotomist (a person trained to draw a blood sample) may be conducted for this purpose. If s/he prefers, s/he may request an in-house clinic visit for the blood draw. In some cases, his/her physician or his/her laboratory may draw his/her blood samples for us. The total amount of blood drawn is about 60 ccs or about 5 tablespoons. This is a small amount and will be rapidly replaced by the body. The blood sample collection visit can be completed in approximately 20 minutes. S/he may also be asked to spit into a vial several times in order to collect DNA from your saliva.

The blood sample will be used to obtain information about the participant's DNA. DNA is the material inside cells that carries genetic information, passing it along to the next generation in structures called

genes. Genes contain information about a person that is inherited from his/her parents, and some of these genes may play a role in our health. We may also examine other laboratory measures associated with longevity (long life) and age-related health conditions.

We plan to use part of the blood sample to examine the participant's cholesterol and other lipid levels and to measure other factors in the blood that may be related to healthy aging. Routine lab tests will be performed once the participant's sample is collected. If any tests fall outside the normal range, the participant (and a physician of his/her choice if s/he desires) will be immediately notified.

We may also ask the participant if we can use part of his/her blood sample to produce a cell line (a family of cells grown in a laboratory). The cell line will continue producing your DNA and can, therefore be used to obtain unlimited amounts of his/her DNA into the future, greatly extending the research usefulness of the LONG LIFE Family Study. By having this cell line, we would be able to study the importance of genes related to longevity that have not even been thought of yet.

All samples will be stored indefinitely at the central site (repository) at the University of Minnesota Medical Center under the direction of Dr. John Eckfeldt. Each specimen will be labeled with a specimen ID number with no personal identifiers. Information linking this code number to the participant's identity will be kept in a separate, locked secure location at the study site and only accessible to study personnel with a different key from that of all other files.

Sharing Your Samples with Other Investigators/Developing Diagnostic & Therapeutic Products

Blood samples may be used by investigators other than the investigators of the current study and will not include information that identifies the participant. We may share his/her sample and anonymous medical information about him/her to other researchers if you, as the informant, and the participant permit us. Samples (blood, DNA or cell lines) may be used to develop one or more diagnostic or therapeutic products which could be patented and licensed. ***PLEASE NOTE: There are no plans to provide any financial compensation to you or the participant should this occur.***

Census Records, Medicare Data and Social Security Number

We will ask you and the participant for his/her social security number for two reasons. First, we will use this social security number to link it to early census records of when s/he was young to ensure that his/her birth date is valid. Since birth date records from many years ago were not always accurate, we want to be absolutely certain that the year in which you believe s/he was born matches the formal records.

From these census records, we will also be able to obtain additional information about his/her siblings, family origin, home ownership, etc, that you may not have been told or may not recall. All of these factors are important because they have been associated with longevity (living to a very old age) in previous studies.

Secondly and with your and the participant's permission, we will use his/her social security number to access data related to his/her health that is maintained by The Centers for Medicare and Medicaid Services (CMS) as well as to access his/her medical records. Since it is often difficult for each of us to remember our complete medical history especially as we grow older and easier to forget what may seem like a minor medical condition, we can best obtain a complete record of his/her medical history using these databases. Since this study is hoping to learn about why people live to a very old age, it is important for us to know as much about his/her medical history as possible to determine how s/he is unique from others who have not aged as successfully as s/he has.

Therefore, his/her social security number will provide us with access to these census and medical data records as this information is saved in national databases under his/her social security number.

Family Member Involvement

You and the participant will also be asked to communicate with other family members to obtain their permission to be contacted by this research group. We will only attempt to contact those family members who agree to participate in the study and they will be invited to undergo similar study procedures as you and the participant.

Telephone Follow-Up

We are requesting that you and the participant continue to complete a short telephone call every twelve months. A staff person will contact you by telephone and update your contact information. This follow up may continue for 10 or more years, until either the study ends or you and the participant chooses to withdraw from the study. This telephone contact will consist of a very brief interview to determine whether the participant has undergone any changes in his/her medical status since we last spoke with you. You will not be asked to come into the clinic. This telephone call will take approximately 10 minutes to complete depending on the number of illnesses and hospitalizations s/he may have had since we last spoke. You will not receive reimbursement for the completion of this brief phone call.

If we are unable to contact you and the participant for this telephone follow-up visit, or you and the participant are either unable to participate or provide consent for the phone interview, we will contact the two close family members you provided us during the in-person visit. One of these family members will be asked the same questions related to the state of the participant's health and whether the participant has had any illnesses or hospitalizations since you and the participant were last contacted.

Audio Taping and/or Videotaping: There will be no audio and/or videotaping involved in this study at any time.

Risks

The Interview and Questionnaires: Your participation as an informant in this study involves minimal risks.

Aside from some boredom or fatigue you may experience during the interviews or the questionnaires, there are no real risks to you. If you and/or the participant becomes tired from the questions, the interview will be stopped at any time and continued another time at your convenience and with the permission of you and the participant. If you, the participant and/or his/her family members are asked questions anyone is uncomfortable answering, any of the questions can remain unanswered at any time. Since we will ask you questions about the health of the participant and his/her family members, there is a risk of disclosure of confidential family matters. We have procedures in place that will protect their confidentiality. These procedures are described below under the Confidentiality Section below.

Risks of the Physical Function Test: Grip strength will be performed on the participant for his/her strongest hand (the hand s/he feels comfortable using most often). This test may result in temporary muscle discomfort, although that is unlikely - occurring less than 10% of the time. There is also a slight risk of the participant losing his/her balance during some of the tests to measure his/her balance and walking speed. The examiner will remain close to help if s/he becomes unsteady.

Risks of the Lung Function Test: Lung function testing, or spirometry, involves minimal risk to the participant. After performing spirometry on nearly 10,000 elders for other research studies, the study investigators have found that, although some participants find the spirometry test ‘tiresome’, there were no serious side-effects. About one in a thousand younger people in workplace settings have fainted during spirometry, but this risk is eliminated by the short (six second) maneuvers that we are using for this particular study. Additionally, since the participant will be performing the test while seated instead of while standing, we have reduced the possibility that s/he may fall if s/he feels faint and/or becomes dizzy.

Risks of the Blood Pressure Testing: After the blood pressure cuff is attached and inflated on the participant’s arm and ankle to measure his/her blood pressure, s/he may experience some mild discomfort with some slight redness once the cuff is removed from either his/her arm or ankle.

Risks During the Blood Draw: You may remember giving a blood sample in the past for tests your doctor wanted to perform. The blood sample that will be drawn from the participant is obtained in a similar fashion using a small needle, and blood is obtained from a vein in his/her. For most people, drawing blood does not cause any serious problems. There is a small risk of bleeding, bruising, discomfort, dizziness, infections and pain at the needle site. However, we will take every precaution using skilled individuals to obtain blood from the participant.

Genetic Information: The Study Investigators may use the participant’s blood sample, including his/her DNA to learn about genes related to longevity and diseases associated with aging, such as heart disease and Alzheimer’s disease. Some of the work to study the participant’s DNA and/or blood sample will also take place in commercial laboratories. However, only the Long Life Family study staff will have access to information that would reveal the participant’s and his/her family members’ identity, such as names, addresses and dates of birth.

This study is looking for similarities in health behaviors, environmental exposures and genes across all enrolled individuals. Therefore, we do not look at a participant’s genes individually. As such, the researchers will not be able to tell the participant how his/her specific results might affect him/her or his/her family members’ future health or other information relating to the participant’s blood samples. If we learn that some genes are linked to people’s future health, lengthy studies will be needed to understand their benefits and risks for any individual person.

There may be unknown risks/discomforts involved. Study staff will provide the participant with timely information on any findings that may affect his/her health, welfare, or decision to remain in this study.

Benefits: This study is not designed for your (as the informant) nor the participant’s benefit, other than knowing that you and the participant may be helping to advance scientific and medical understanding. Your and the participant’s participation may lead to findings that benefit people, through a better understanding of aging and how to reduce its problems and diseases. It typically takes many years to translate such findings into helpful strategies and therapies.

Compensation: There will be no costs to you or to the participant for participation in this study.

Confidentiality: Any information about you and the participant obtained from this research study will be kept as confidential (private) as possible. All records related to your and the participant’s involvement in this research study will be stored in locked file cabinets. Your and the participant’s identity on these records will be indicated by a case number rather than your and the participant’s name, and the information linking these case numbers with your/his/her identity will be kept separate from the research records. Your and the participant’s identity and participation in this study will be maintained in a confidential database at the Data

Coordinating Center at Washington University in St. Louis, Missouri. All records will be stored in a computer file and access will be restricted to research staff. Each blood specimen will be labeled with a specimen ID number without any information that identifies you or the participant. Information linking this code number to your identity will be kept in a separate, locked secure location at the Field Center and only accessible to study personnel with a different key from that of all other files. You will not be identified by name in any publication of the research results unless you sign a separate form giving your permission (release).

To help us protect your and the participant's privacy, we have obtained a Certificate of Confidentiality from the National Institutes of Health. With this Certificate, the researchers cannot be forced to disclose information that may identify you or the participant, even by a court subpoena, in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings. The researchers will use the Certificate to resist any demands for information that would identify you or the participant, except as explained below.

The Certificate cannot be used to resist a demand for information from personnel of the United States Government that is used for auditing or evaluation of Federally funded projects or for information that must be disclosed in order to meet the requirements of the federal Food and Drug Administration (FDA).

The Certificate of Confidentiality does not prevent the researchers from disclosing voluntarily, without your or the participant's consent, information that would identify you or the participant in the research project under the following circumstances. However, if the researchers learn that you or the participant are in real danger of physical or serious mental harm (example, suspected or known sexual or physical abuse of a child or threatened violence to self or others) they will release study related information to protect you, the participant and the other persons. Such information must be reported to the appropriate authorities.

You and the participant should also understand that this Confidentiality Certificate does not prevent you, the participant or a member of your/his/her family from voluntarily releasing information about you/the participant or your/his/her participation in this research. If an insurance company, employer or other employer obtains your/the participant's written consent to receive research information, the Certificate of Confidentiality will not protect his/her privacy.

Research Standards and Rights of Participants: Participation in this research study is voluntary. If you or the participant decide not to participate, or if you or the participant later decide to stop participating, you or the participant will not lose any benefits to which you or the participant are otherwise entitled. A decision not to participate will not effect your or the participant's treatment at the New York Psychiatric Institute. Additionally, your or the participant's decision to participate or not participate will have no effect on your or the participant's Medicare benefits. All information you and the other participants provide is protected under the Privacy Act and the Health Insurance Portability and Accountability Act.

You and the participant will be notified of significant new findings that may occur during the course of the study and that may relate to your or the participant's willingness to continue to participate.

Federal regulations require that research participants be informed about our institutions' policies with regard to the provision of treatment and compensation for research related injuries. If you or the participant believe that you or the participant have sustained injury as a result of participating in a research study, you or the participant may contact the Principal Investigator, Dr. [REDACTED] at [insert appropriate phone number], or the Administrative Director of the [insert State name] State Psychiatric Institute Institutional Review Board at [REDACTED], so that you or the participant can review the matter and identify the medical resources which may be available to you or the participant.

Please be aware that:

1. The [insert State name/Institution name] will furnish that emergency medical care determined to be necessary by the medical staff of this hospital
2. You or the participant will be responsible for the cost of such care, either personally or through your or the participant's medical insurance or other form of medical coverage.
3. No monetary compensation for wages lost as a result of injury will be paid to you or the participant by the [insert State name/Institution name].
4. By signing this consent form, you are not waiving any of your legal rights to seek compensation through the courts.

The investigator will answer to the best of his ability any questions that you may have now or in the future about the procedures or about your response to the procedures. You may reach Dr. [REDACTED], principal investigatory, by calling [insert appropriate phone number].

The [insert State name/Institution name] Review Board has approved the recruitment of subjects for this study. If you have any questions about your rights as a research subject or any complaints, you may call the Administrative Director at [insert appropriate phone number] during regular office hours.

You will be given a copy of the Consent Form to keep.

**Give
Consent**

**Don't Give
Consent**

Will you or the participant allow us to measure cholesterol and other blood factors and release any findings from these tests to the participant's physician?

Will you or the participant allow us to prepare and test DNA from the participant's blood cells for genes that may be related to age-related health conditions as well as for those genes related to other conditions not age-related?

Will you or the participant us to create and store a cell line at the repository at the University of Minnesota Medical Center?

Will you or the participant allow us to share the participant's blood samples with other researchers who are not part of this study who may use these samples for other types of research?

Will you or the participant allow us to release findings from study-related tests and examinations to the participant's physician?

Documentation of Consent:

Consent by Participant: I have read the above and understand the risk, benefits and alternatives (including non-participation). I voluntarily agree to participate in the research study described above.

Signature of Participant (Informant)

Date

PLEASE PRINT: Name of Participant (Informant)

I have discussed the proposed research with this participant, and, in my opinion, this participant understands the benefits, risks and alternatives (including non-participation) and is capable of freely consenting to participate in research.

Signature of Person Obtaining Consent

Date

The LONG LIFE Family Study

LLFS



Progeny 6 Tutorials for LLFS

Version 1.0 - September 27, 2007


LLFS Data Management Coordinate Center
Department of Genetics - Division of Statistical Genomics
Washington University School of Medicine
4444 Forest Park Blvd., Campus Box 8506
St. Louis, MO 63110-1093
Phone: (314) 362-3645 Fax: (314) 362-2693
e-mail: asleeter@dsgmail.wustl.edu

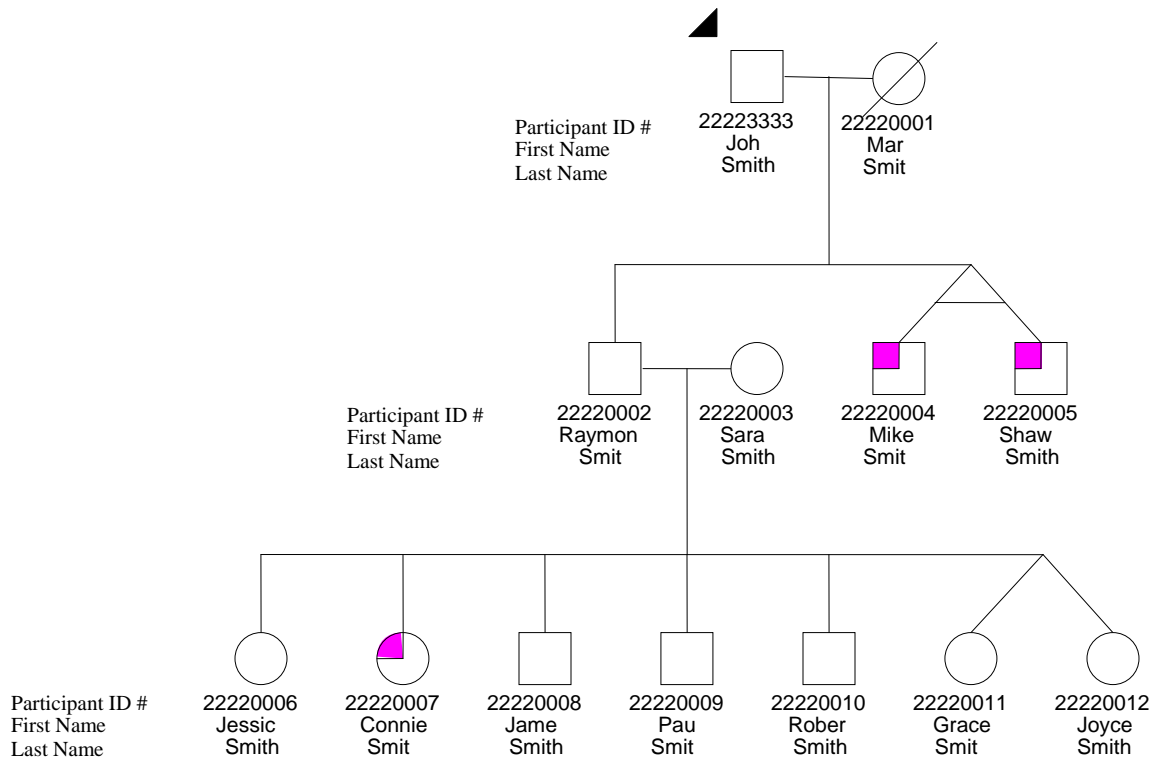
Progeny 6 Tutorials for LLFS

This tutorial is developed mainly for drawing pedigrees, and further exporting the pedigree structures into a triplet data file for the Long Life Family Study (LLFS). It includes instructions of drawing a pedigree, entering individual data, and exporting each individual's triplet information (participant ID, father ID, and mother ID) into a text file. Below are a sample plot of a three-generation pedigree and a table of each individual's triplet data. Following the steps described in this document, you will be able to draw the sample pedigree and generate the sample triplet data as well.

Sample Pedigree Plot:

Pedigree Name - 22223333

 Smoke Now = 1(Yes)



Sample triplet data:

Folder Name	Pedigree Name	UPN *	Patient ID #	Father ID **	Mother ID **	Gender	Proband status	Twin Relationship	Twin status
LLFS	22223333	1	22223333	0	0	M	1		0
LLFS	22223333	3	22220001	0	0	F	0		0
LLFS	22223333	5	22220002	1	3	M	0		0
LLFS	22223333	8	22220006	5	15	F	0		0
LLFS	22223333	9	22220007	5	15	F	0		0
LLFS	22223333	10	22220008	5	15	M	0		0
LLFS	22223333	12	22220010	5	15	M	0		0
LLFS	22223333	15	22220003	0	0	F	0		0
LLFS	22223333	17	22220004	1	3	M	0	Monozygotic	1
LLFS	22223333	20	22220005	1	3	M	0	Monozygotic	1
LLFS	22223333	21	22220011	5	15	F	0	Dizygotic	1
LLFS	22223333	22	22220012	5	15	F	0	Dizygotic	1
LLFS	22223333	23	22220009	5	15	M	0		0

*UPN: Unique # of individual in a pedigree. UPN is automatically generated by the software.

** Father ID / Mother ID: Defines UPN of father/mother of an individual.

Each Field Center will draw the pedigrees based on individuals' genetic relationship, not on the social relationship, and create a triplet file that contains each individual's triplet information for all individuals in each pedigree. **Please note that the triplet file (in .csv format) should have the exactly same columns as the above sample table.**

This document serves as the user guide for LLFS and describes the basic methods for the following aspects:

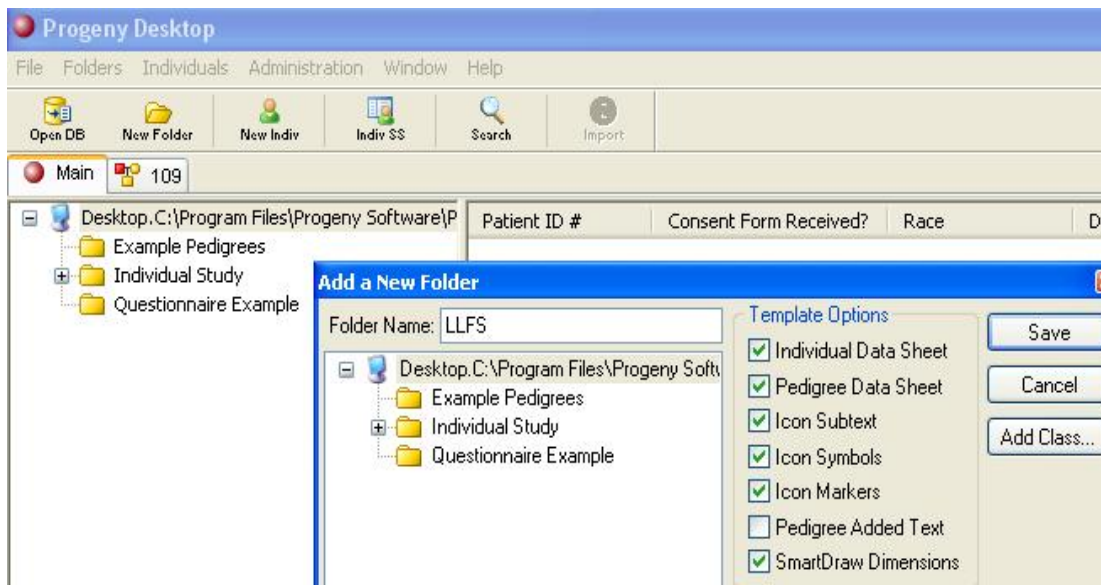
1. Creating New Folder "LLFS" to Organize Pedigrees and Individuals
2. Create and Draw Pedigrees
3. Formatting Pedigree Icon Subtext
4. Entering Pedigree and Individual Data
5. Querying and Exporting Pedigree and Individual Data

The Progeny 6 functions like a standard database and has a graphical user interface, allowing you to create database fields for pedigrees and fields for individuals. It also allows customizations that help you see how all features of recruitment are processed. If you are interested in learning how to perform more functions in Progeny 6, you may click the Help on the menu bar and choose User Manual, or view Progeny 6 Online video demonstrations on the website: <http://www.progenygenetics.com/tutorials.html>.

1. Creating New Folder "LLFS" to Organize Pedigrees and Individuals

- To create the "LLFS" folder, first click the **Pedigrees** bar on the bottom left navigation pane of the Main Screen.
- To create a new folder under the root directory, click/highlight the root (it is the location the software was installed on your computer, for example, Desktop.C:\ ... \Progeny6Demo.db.)

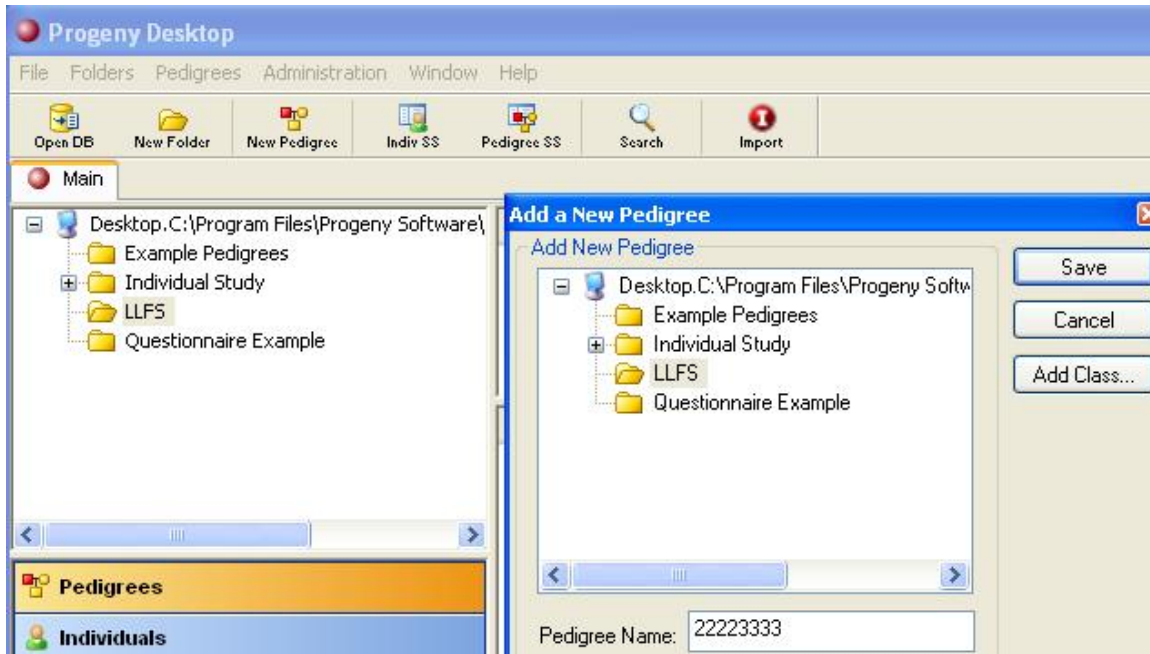
- Click the **New Folder** button on the toolbar (you may also right-click in the Pedigree Folders window, or choose the menu option Folders and select **New Folder**.) The "Add a New Folder" screen will appear (see the figure below).
- Enter "**LLFS**" without the quote in the Folder Name for the new folder.
- In the **Template Options** box, clicking options Individual Data Sheet, Pedigree Data Sheet, Icon Symbols, Icon Subtext, and SmartDraw Dimensions will have all the pedigrees within a folder remain consistent fields.
- Click **Save** and the new folder LLFS will be created under the root directory. Now, you can begin to enter all the pedigrees in LLFS for the folder.



2. Create and Draw Pedigrees

2.1. Create a pedigree

- To create a new pedigree, first click the **Pedigree** bar in the navigation pane, and click the **New Pedigree** button on the toolbar or right-click inside the pedigree listings and choose **New Pedigree**.
- Choose the **LLFS** folder, and then enter a name for the pedigree. **PLEASE NOTE: THE NAME OF A PEDIGREE IS THE 8-DIGIT PROBAND ID OF A PEDIGREE IN LLFS.** (Tip: To avoid any possible data entry error, you may copy a proband ID from an existing file and paste it as the name of the pedigree.)



- Once the pedigree is saved, it will appear as an icon on the right side of the screen. Double-click the pedigree icon to launch the Pedigree drawing screen.

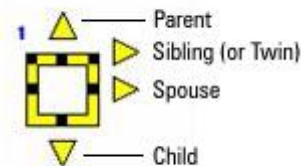
2.2. Draw a pedigree

Typically, in a pedigree diagram, males are identified as square icons, females are circle icons, and sex-unknowns are depicted as diamonds. The lines connecting individuals identify the status of relationships between two individuals, such as marital status.

- There are three ways to add individuals to a pedigree:
 - (1) Right-clicking
 - (2) One-Click Add
 - (3) Palette

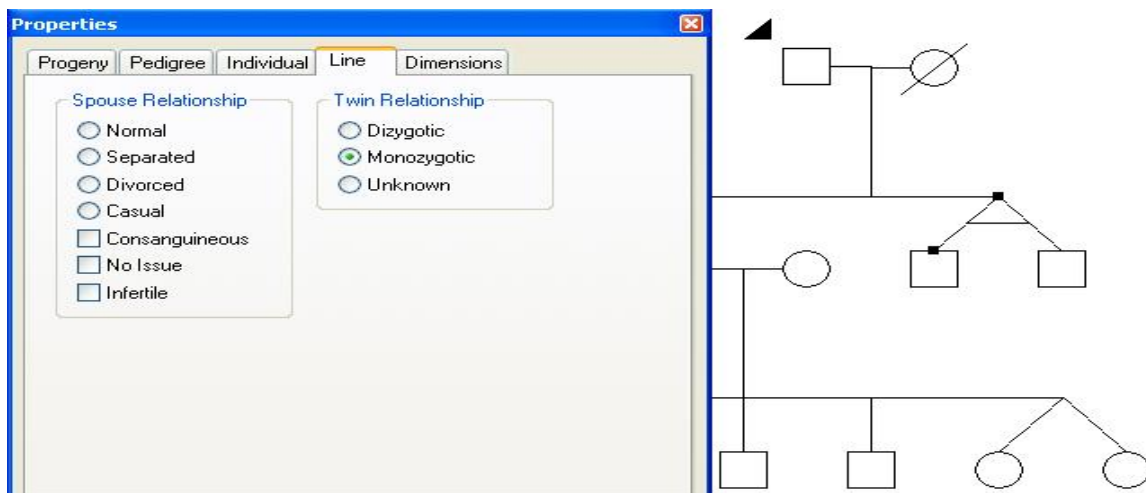
The One-click Add option is the fastest way, and we'll here use the One-click Add to add individuals to the pedigree.

- To select the checkbox **One-click Add**, open the **Properties** screen by right-clicking on the canvas of the pedigree and select **Properties**. Check the checkbox One-click Add that is under the **Progeny Tab** in the **Properties** screen.
- By selecting the One-click Add option, each time you highlight an individual in the pedigree, yellow arrows (see the figure right) will appear on all sides of the individual.
- To add the first individual, right-click on the canvas, and choose **Add Individual** and then further choose either Add Male or Add Female. Click on this individual and use yellow arrows to add more individuals within the pedigree.
- To add a male, use the **left** mouse button and click on the arrow. To add a female, click on the **right** mouse button. Each time you click on the yellow arrow, it will add that relation to the individual.



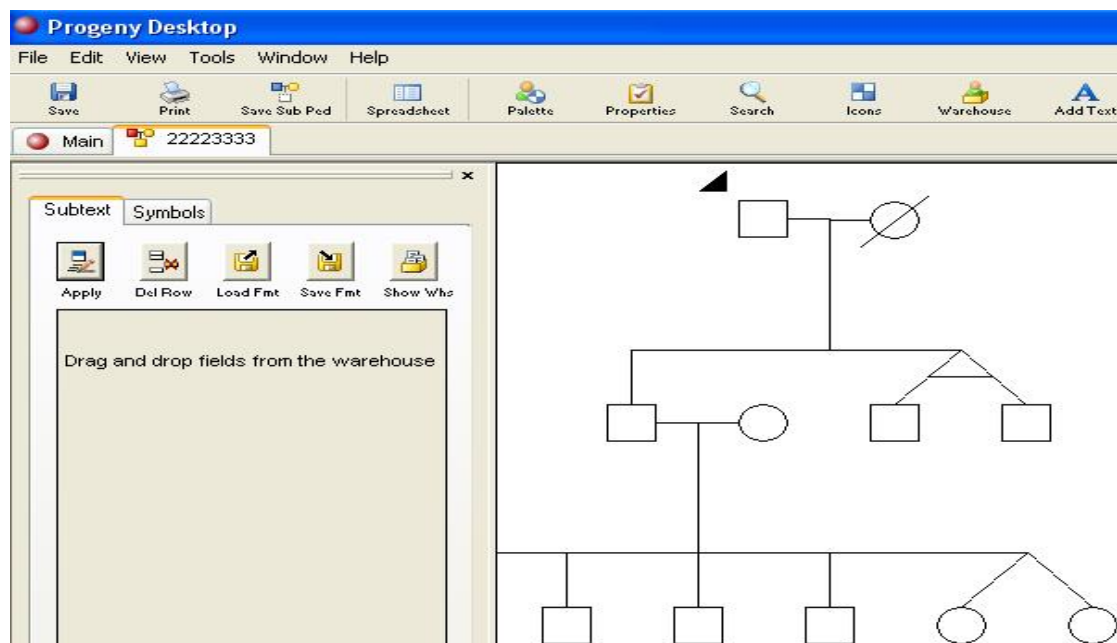
Note (Add a Twin):

- To add a twin, hold **CTRL** key while clicking the sibling yellow arrow.
- To add a twin brother, click the left mouse button, and add a twin sister, click the right mouse button.
- To distinguish monozygotic or dizygotic twin relationship, click on the line angling up from one of the twins, open the **Properties** window, and choose it to monozygotic, dizygotic, or unknown under **Line** tab.
- The following figure shows the Properties window, and a male monozygotic twin and a female dizygotic twin in a pedigree plot.

**3. Formatting Pedigree Icon Subtext**

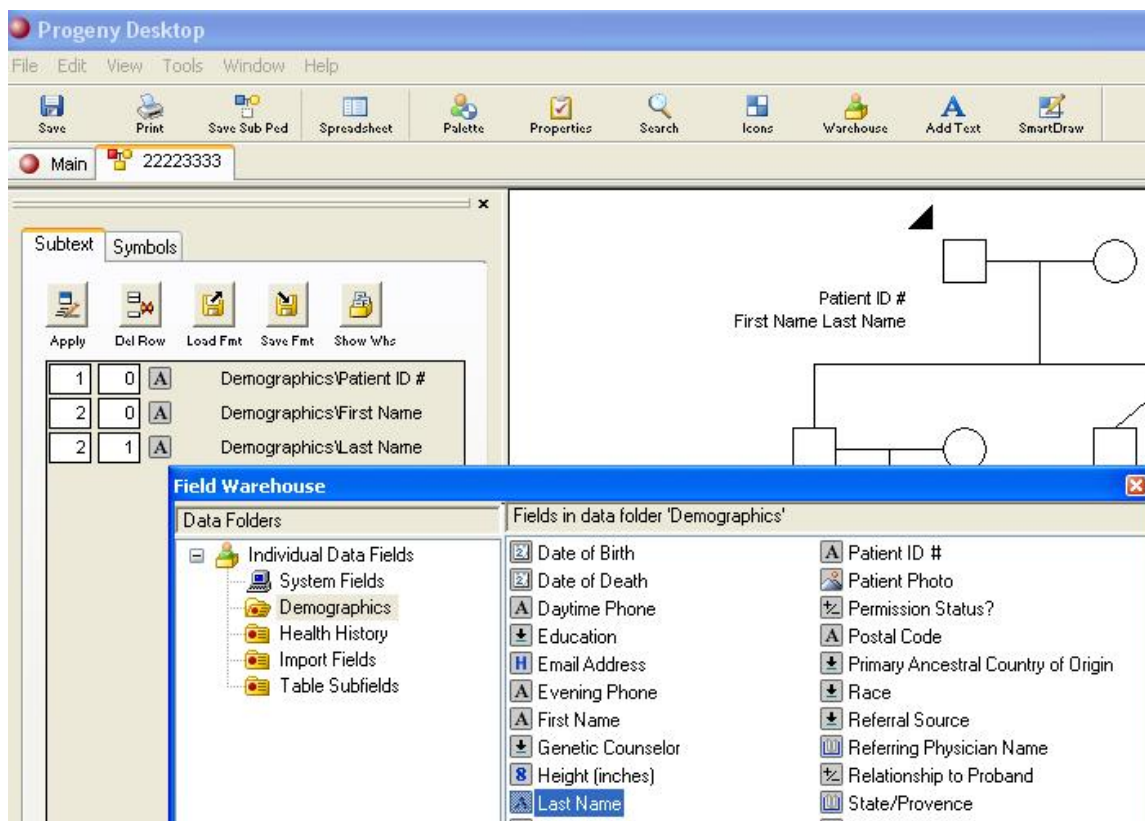
In a pedigree plot, information under an icon is specific to that individual and is called Icon Subtext. The followings describe how to choose and format icon subtext.

- Click the **Icons** button on the toolbar to open the Icons window. The Icons window will be opened at the left side of the screen as follows.



- Click the **Warehouse** button on the toolbar to open the Warehouse Field window, and highlight the Demographics folder under the Data Folders.
- Drag and drop the fields you want to appear under the icon to the Icons window. **Please make sure that you drag and drop the field **Patient ID #** because this field will be used for entering/displaying LLFS participant ID.** You may also want to select First Name, Last Name, or any other fields that will be displayed as the icon subtext.
- You can place fields side by side under an icon. The second column of the **Subtext** Tab in the left top of the Icons window (see the following figure) is for determining the positioning of the text fields within the same row.
- To remove fields from the Icon Subtext tab, highlight the field and click the **Del Row** button or right-click on the field and choose **Delete Selected Row**.
- Close the Field Warehouse window and click **Apply** when you are finished.

Note: The options **Save Fmt** and **Load Fmt** can be used to save/use different formats for icon subtext. This is ideal for displaying different sets of data instantly, without having to re-select the fields you want to display. The **Save Fmt** button and **Load Fmt** button are located on the top of the Icons window.

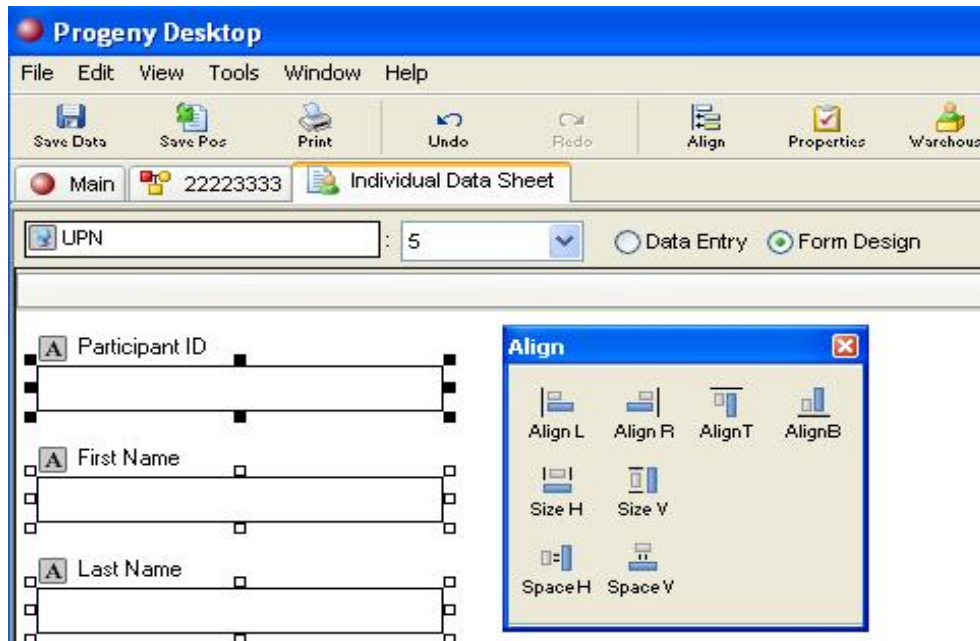


4. Entering Pedigree and Individual Data

4.1. Individual Data Sheet

The Individual Data Sheet is the data entry form for individuals in the database. It can be customized to look like a form or a questionnaire. By using the Template Option within a folder (see section 1. Creating New Folder "LLFS"), each individual will have the same exact Individual Data Sheet design within the same folder. Changes made to one Individual Data Sheet will be reflected in all the data sheets within the Folder.

- To launch the Individual Data Sheet, double-click an individual icon in a pedigree.
- To format an Individual Data Sheet, first, click the **Form Design** radio button.
- Next, click Warehouse button on the toolbar, and drag and drop the fields: Patient ID #, First Name, Last Name, or any other fields to the canvas (**Please Note: you may drag and drop as many fields as you want, but the field Patient ID # is required for later entering the LLFS participant ID.**) Close the Field Warehouse window after you drag and drop the fields.
- Click the **Align Palette** in the toolbar and use it to assist you in lining up fields on the form, re-sizing a field, or spacing between fields. Hold down the **CTRL** key, select multiple fields, and click the button for the desired action, for example, clicking the button AlignL aligns the left edges of the selected fields vertically. Also, groups of fields can be moved simultaneously by holding down the **CTRL** key (see the figure below).



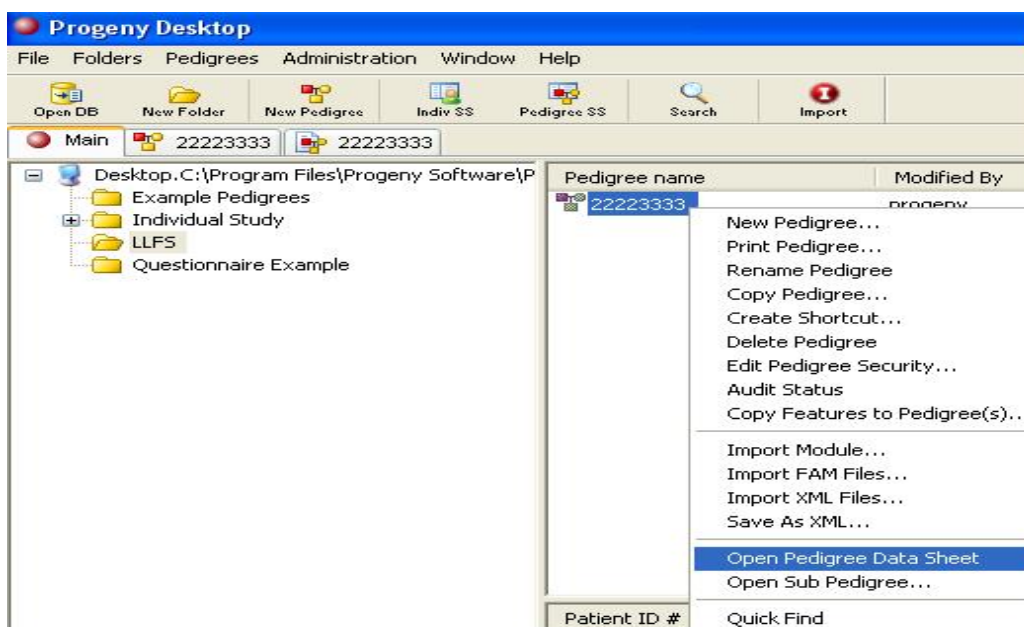
- When you are finished setting up the Individual Data Sheet, click the **Data Entry** radio button, click the "x" on the top of the right corner of the Individual Data Sheet window to close the window, and then click the **Save** button.
- Now, you are ready to enter data for each individual. Double-click an individual icon on the canvas to open the Individual Data Sheet, and then enter data for the individual. (**Tip: To avoid any possible data entry error, you may copy each individual's LLFS ID or other data from an existing file and paste them into each filed.**) Repeat this step till the data for all individuals within a pedigree are entered.

4.2. Pedigree Data Sheet (optional)

The Pedigree Data Sheet provides a method to enter data for an entire pedigree. Pedigree Data Sheets are unique per Pedigree Folder. All pedigrees within a single folder will have the same Pedigree Data Sheet if you choose the Template Option to allow for this.

This step is optional for LLFS. However, if you want to know which date the pedigree was created, which date was last modified, and who modified it, etc., the Pedigree Data Sheet is a good way to keep track down all those information.

- The way to set up the Pedigree Data Sheet is similar to the way for the Individual Data Sheet. To launch the Pedigree Data Sheet, right-click on a pedigree name and then select **Open Pedigree Data Sheet** to open the following Pedigree Data Sheet window.
- To format a Pedigree Data Sheet, first, click the **Form Design** radio button located at the top of the Pedigree Data Sheet window.



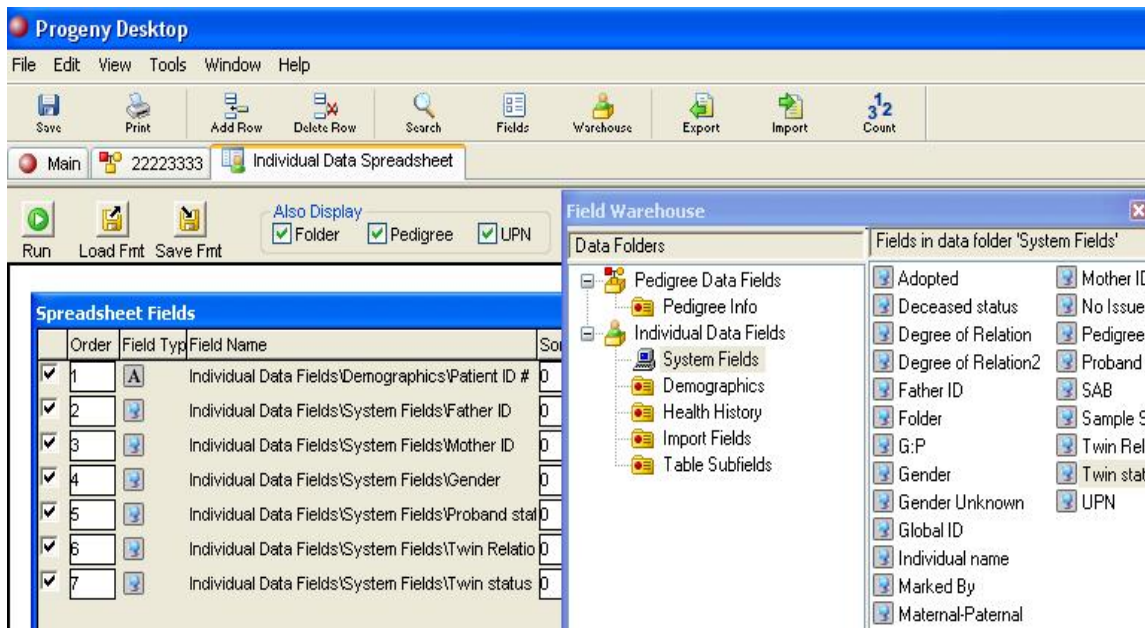
- Next, click the button **Warehouse** on the toolbar, highlight Pedigree Info folder, and drag and drop the fields you want to the canvas. (Note: you may create a new field if it is needed. The instructions for creating a new field can be found in the Progeny 6 User Manual.)
- You can use the **Align Palette** to align up the fields in the window (Details can be found in section 4.1.)
- Click the **Data Entry** radio button, close the Pedigree Data Sheet window and click the **Save** button when you are finished. Now, you are ready to enter the data for each pedigree.

5. Querying and Exporting Pedigree and Individual Data

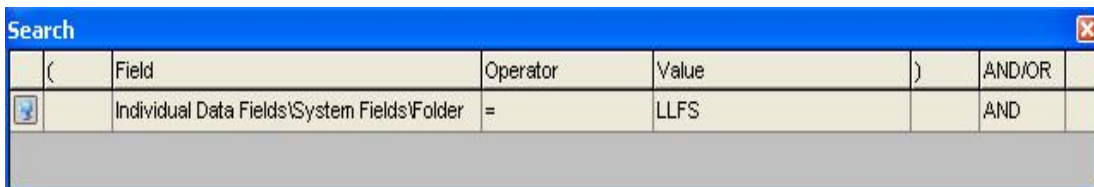
Querying Pedigree and Individual Data

To obtain each individual's triplet data, you can run an Individual Spreadsheet for the entire database, and then query the spreadsheet to only include individuals in the pedigrees from the particular folder LLFS.

- To launch the Individual Spreadsheet, click the "**Indiv SS**" button on the toolbar in the Main Screen. The Spreadsheet Fields window and the Field Warehouse window will appear simultaneously.
- Drag and drop the following seven fields from the Field Warehouse window to the Spreadsheet Fields window (**Note: All the fields can be found in the System Filed folder, except the Patient ID # in the Demographics folder**):
 - Patient ID #
 - Father ID
 - Mother ID
 - Gender
 - Proband Status
 - Twin Relationship
 - Twin Status



- Check the check boxes: **Folder, Pedigree, and UPN** on the top of the Individual Spreadsheet window.
- After setting up the Spreadsheet Fields, click **Run** button and a large spreadsheet will be generated.
- The next step will be querying the spreadsheet by clicking the **Search** button on the toolbar. Again, drag and drop the Folder field in the System Filed folder from the Warehouse window to the following window, and then enter LLFS in the Value column.

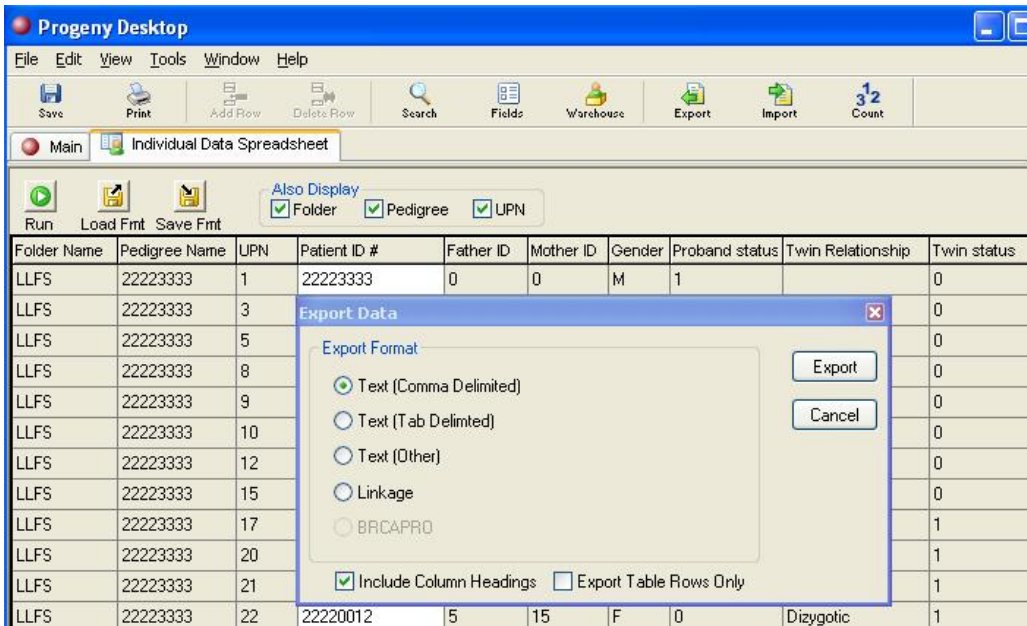


- Click the **Run** button, and the spreadsheet will be narrowed down to only include individuals in the pedigrees within the LLFS folder. This is the spreadsheet that contains the triplet data for LLFS.

Note: You can save and recall the frequently run searches. Once the spreadsheet is generated, click the **Save Fmt** button and name the format. You can recall this search by clicking the **Load Fmt** button and choosing the name of the saved search, then clicking **Run**.

Exporting Pedigree and Individual triplet Data

- Once the spreadsheet with triplet data is generated, click the **Export** button on the toolbar. The following window will appear:



- Choose the radio button **Text (Common Delimited)**, and check the check box **Include Column Headings** in the Export Data window, and then click **Export** button.
- Save the triplet file with a File Name: **triplet_XX_MMDDYYYY**, where XX is a two-character center code for each field center, i.e. BU for Boston, NY for Columbia, and PT for Pittsburgh, and MMDDYYYY is the date of creating the triplet file, for example, 09152007 for September 15, 2007.

If you have any questions regarding the tutorials, please e-mail Rosa Lin at rosa@wustl.edu.



Appendix D

Data Entry Systems Users' Guide

The Data Entry Systems Users' Guide is not yet available; this will be provided at the time of training.



Appendix F

LLFS Collaborators List/Directory

Field Center/Affiliation	Name, Address, Etc.	Contact Information
Project Office NIA / NIH	Hadley, Evan C. MD Associate Director, GCGP Gateway Bldg, Ste. 3C307 7201 Wisconsin Avenue Bethesda, MD 20892-9205	Ph: (301) 435-3044 Fax: (301) 402-1784 EM: ehadley@nih.gov
Project Office NIA / NIH	Dutta, Chhanda PhD Chief, Clinical Gerontology Branch Gateway Bldg, Ste. 3C-307 7201 Wisconsin Ave Bethesda, MD 20892-9205	Ph: (301) 435-3048 Fax: (301) 402-1784 EM: duttac@nia.nih.gov
Project Office NIA / NIH	Patmios, Georgeanne E. Acting Branch Chief, Population & Social Processes Branch, BSRP Gateway Bldg, 7201 Wisconsin Ave., Ste. 533 Bethesda, MD 20892-9205	Ph: (301) 496-3138 Fax: (301) 402-0051 EM: patmiosg@nia.nih.gov
Project Office NIA / NIH	Simonsick, Eleanor 3001 S. Hanover Street Baltimore, MD 21225	Ph: (410) 350-3965 Fax: (410) 350-7304 EM: simonsickel@mail.nih.gov or es37m@nih.gov
Project Office NIA / NIH	Suzman, Richard PhD Associate Director, BSRP Gateway Bldg., 7201 Wisconsin Ave., Ste. 533 Bethesda, MD 20892-9205	Ph: (301) 496-3138 Fax: (301) 402-0051 EM: suzmanr@nia.nih.gov
Project Office NIA / NIH	Rossi, Winifred K. MA Deputy Assoc. Director, GCGP 7201 Wisconsin Ave, Ste. 3C307 Bethesda, MD 20892-9205	Ph: (301) 496-3836 Fax: (301) 402-1784 EM: rossiw@nia.nih.gov or Winnie_rossi@nih.gov
DMCC / Washington University School of Medicine	Province, Michael A. PhD (PI) Professor of Genetics and Biostatistics Division of Statistical Genomics 4444 Forest Park, Campus Box 8506 St. Louis, MO 63108	Ph: (314) 362-3616 Fax: (314) 362-4227 EM: mprovince@wustl.edu
DMCC / Washington University School of Medicine	Borecki, Ingrid PhD Assoc. Professor of Genetics/Biostat Division of Statistical Genomics 4444 Forest Park, Campus Box 8506 St. Louis, MO 63108	Ph: (314) 362-3690 Fax: (314) 362-4227 EM: iborecki@wustl.edu
DMCC / Washington University School of Medicine	Corbett, Jonathan PhD Research Instructor Division of Statistical Genomics 4444 Forest Park, Campus Box 8506 St. Louis, MO 63108	Ph: (314) 362-362-3593 Fax: (314) 362-4227 EM: jon@wustl.edu
DMCC / Washington University School of Medicine	Feitosa, Mary PhD Research Asst. Professor Division of Statistical Genomics 4444 Forest Park, Campus Box 8506 St. Louis, MO 63108	Ph: (314) 747-3792 Fax: (314) 362-4227 EM: maryf@wustl.edu
DMCC / Washington University School of Medicine	Miller, J. Philip AB Professor of Biostatistics 660 South Euclid, Box 8067 St. Louis, MO 63110	Ph: (314) 362-3617 Fax: (314) 362-3728 EM: phil@wubios.wustl.edu

Field Center/Affiliation	Name, Address, Etc.	Contact Information
DMCC / Washington University School of Medicine	Morgan, Derek AB Database Administrator Division of Statistical Genomics 4444 Forest Park, Campus Box 8506 St. Louis, MO 63108	Ph: (314) 362-3685 Fax: (314) 362-4227 EM: derek@wustl.edu
DMCC / Washington University School of Medicine	Sleeter, Amy J. BS Project Manager Division of Statistical Genomics 4444 Forest Park, Campus Box 8506 St. Louis, MO 63108	Ph: (314) 362-3945 Fax: (314) 362-4227 EM: asleeter@wustl.edu
DMCC / Washington University School of Medicine	Valentine, Jim PhD Senior Statistical Data Analyst Division of Statistical Genomics 4444 Forest Park, Campus Box 8506 St. Louis, MO 63108	Ph: (314) 362-3688 Fax: (314) 362-4227 EM: jimv@wustl.edu
DMCC / Washington University School of Medicine	Adelman, Avril, Sr. Stat Data Analyst Lin, Rosa, Statistical Data Analyst Wu, Jun, Research Statistician	EM: avril@wustl.edu ; (314) 362-2445 EM: rosa@wustl.edu ; (314) 362-3732 EM: jwu@wustl.edu ; (314) 362-3940
Boston University / Boston Medical Center	Perls, Thomas MD, MPH (PI) Geriatrics, Robinson 2400 88 East Newton Street Boston, MA 02118	Ph: (617) 638-6688 (Alt: 617-733-7893) Fax: (617) 638-6671 EM: thperls@bu.edu
Boston University / Boston Medical Center	Ash, Arlene PhD School of Public Health 715 Albany Street Boston, MA 02118	Ph: (617) 638-7518 Fax: EM: aash@bu.edu
Boston University / Boston Medical Center (Subcontract)	Elo, Irma PhD Population Studies Center University of Pennsylvania 239 McNeil, 3718 Locust Walk Philadelphia, PA 19104-6298	Ph: (215) 898-6441/9162 Fax: (215) 898-2124 EM: popelo@pop.upenn.edu
Boston University / Boston Medical Center	Sebastiani, Paolo PhD Associate Professor Department of Biostatistics 426 E Talbot Bldg, 715 Albany St. Boston, MA 02118	Ph: (617) 638-5877 Fax: (617) 638-6484 EM: sebas@bu.edu
Boston University / Boston Medical Center	Terry, Lara MD, MPH Geriatrics, Robinson 2400 88 East Newton Street Boston, MA 02118	Ph: (617) 638-6688 Fax: (617) 638-6671 EM: laterry@bu.edu
Boston University / Boston Medical Center	Anderson, Stacy, Site Manager McLaughlin, Kirsten, Research Asst. Pennington, JaeMi, Research Asst. Wilson, Cherrell, Research Asst.	EM: stacy@bu.edu ; Ph: (617) 638-6679 EM: kirstenm@bu.edu ; EM: jaemi@bu.edu ; Ph: (617) 638-6683 EM: cherrell@bu.edu ; Ph: (617) 638-6680
Central Labs, University of Minnesota	Eckfeldt, John H. MD, PhD Professor & Vice Chair, Clinical Affairs Dept. Lab. Med./Pathology, MMC 609 Univ. of MN, Room 763-1 Mayo Bldg. 420 Delaware Street, SE Minneapolis, MN 55455	Ph: (612) 626-3176 Fax: (612) 626-2696 EM: eckfe001@umn.edu
Central Labs, University of Minnesota	Foster, Cathie Leiendecker MS, CLSp Dept. Lab. Med./Pathology University of Minnesota Mayo Mail Code 609, Rm 763-C Mayo 420 Delaware Street, SE Minneapolis, MN 55455	Ph: (612) 626-2305 Fax: (612) 626-2696 EM: foste011@umn.edu

Field Center/Affiliation	Name, Address, Etc.	Contact Information
Central Labs, University of Minneosta (Fairview Subcontract)	Arends, Valerie Rynders, Greg	EM: varends1@Fairview.org EM: grynder1@fairview.org
Columbia University-New York	Mayeux, Richard MD, MSc (PI) Sergievsky Prof of Neurology, Psychiatry, Epidemiology Co-Director, Taub Institute of Research College of Physicians & Surgeons 630 West 168 th Street New York, New York 10032	Ph: (212) 305-2391 Fax: (212) 305-2518 EM: rpm2@columbia.edu Assistant: Mary Anne Barsamian EM: msb@sergievsky.cpmc.columbia.edu
Columbia University-New York	Costa, Rosann MA Senior Research Associate College of Physicians & Surgeons 630 West 168 th Street Ph 19 – Room 304 New York, New York 10032	Ph: (212) 305-9346 Fax: (212) 305-2426 EM: rc181@columbia.edu
Columbia University-New York	Lee, Joseph H. DrPH Assistant Professor of Epidemiology Sergievsky Center College of Physicians and Surgeons 630 West 168th Street New York, New York 10032	Ph: (212) 305-6022 Fax: (212) 305-2426 EM: jhl2@columbia.edu
Columbia University-New York	Schupf, Nicole PhD, MPH, DrPH Assoc. Professor, Clinical Epidemiology Taub Institute for Research College of Physicians & Surgeons P.O. Box 16, 630 West 168th Street New York, NY 10032	Ph: (212) 305-2381 Fax: (212) 305-2426 EM: ns24@columbia.edu
Columbia University-New York	Cosentino, Stephanie Stern, Yaakov Bloom, Jennifer , Research Asst. Cheringal, Julia , Research Asst. Lopez, Yilliam , Research Asst. Sangobowale, Mike , Research Asst.	EM: scosentino@sergievsky.cpmc.columbia.edu EM: ys11@columbia.edu EM: jbloom@sergievsky.cpmc.columbia.edu EM: jcheringal@sergievsky.cpmc.columbia.edu EM: ylopez@sergievsky.cpmc.columbia.edu EM: msangobowale@sergievsky.cpmc.columbia.edu
University of Southern Denmark / Duke University	Christensen, Kaare MD, PhD, DMsc Professor of Epidemiology Director of Danish Aging Research Ctr. Campusvej 55 DK-5230 Odense M Denmark	Ph: +45 6550 3049 (+45 6550 3029) Fax: +45 6550 3682 EM: kchristensen@health.sdu.dk
University of Southern Denmark / Duke University	Yashin, Anatoli Ph.D. Center for Demographic Studies Duke University 2117 Campus Drive, Box 90408 Durham, NC 27708-0408	Ph: (919) 668-2713 Fax: EM: yashin@cds.duke.edu
University of Southern Denmark / Duke University	Bodker, Frans Jakob Mortensen Christiansen, Lene	EM: fbodker@health.sdu.dk EM: jmortensen@health.sdu.dk EM: lchristiansen@health.sdu.dk

Field Center/Affiliation	Name, Address, Etc.	Contact Information
University of Pittsburgh	Newman, Anne B. MD, MPH (PI) Professor of Epidemiology & Medicine Healthy Aging Research Prog. Bellefield Professional Building 130 North Bellefield Avenue-Room 532 Pittsburgh, PA 15213	Ph: (412) 383-1871 Fax: (412) 383-1308 EM: newmana@edc.pitt.edu Assistant: Donna Adams Ph: (412) 383-1931 Fax: (412) 383-1308 EM: adamsd@edc.pitt.edu
University of Pittsburgh	Barmada, M. Michael Ph.D. Asst. Professor, Human Genetics Graduate School of Public Health 130 Desoto St., Rm A300 Crabtree Hall Pittsburgh, PA 15261	Ph: (412) 383-7959 Fax: (412) 624-3020 EM: michael.barmada@hgen.pitt.edu
University of Pittsburgh	Glynn, Nancy W. PhD Assistant Professor of Epidemiology University of Pittsburgh Department of Epidemiology Bellefield Professional Building 130 N. Bellefield Ave., Room 507 Pittsburgh, PA 15261	Ph: (412) 383-1309 Fax: (412) 383-1308 EM: glynnn@edc.pitt.edu
University of Pittsburgh	Kammerer, Candace M. PhD Assoc. Professor, Human Genetics Graduate School of Public Health 130 DeSoto St., Rm A310A Crabtree Hall Pittsburgh, PA 15261	Ph: (412) 624-7265 Fax: (412) 624-3020 EM: ckammerer@hgen.pitt.edu
University of Pittsburgh	Zmuda, Joseph PhD Asst. Professor of Epidemiology Graduate School of Public Health 130 DeSoto St., Rm A544 Crabtree Hall Pittsburgh, PA 15261	Ph: (412) 624-2970 Fax: (412) 624-7397 EM: zmudaj@edc.pitt.edu
University of Pittsburgh	Kost, Piera , Trainer Taylor, Christopher , Data Entry Davis, Latisha , Data Entry	EM: kostp@edc.pitt.edu , (412) 624-2914 EM: cat16@pitt.edu , (412) 383-1871 EM: davisl@edc.pitt.edu
University of Pittsburgh / Johns-Hopkins Subcontract	Fallin, Dani Assistant Professor Johns Hopkins University Bloomberg School of Public Health Dept. of Epidemiology Wolfe Street, #6509 Baltimore, MD 21224	Ph: (410) 955-3463 Fax: (410) 955-0863 EM: dfallin@jhsph.edu
University of Pittsburgh / Johns-Hopkins Subcontract	Walston, Jeremy MD Johns Hopkins Medical Institutions Bayview Medical Center, Johns Hopkins Bayview Care Center 5505 Hopkins Bayview Circle Baltimore, MD 21224	Ph: (410) 550-1003 Fax: (410) 550-2116 EM: jwalston@jhmi.edu
Consultant / CMS	Duzor, Spike Centers for Medicare/Medicaid Svcs Office of Research, Dev & Info Room C3-16-07, Center Building 7500 Security Blvd. Baltimore, MD 21244-1850	Ph: (410) 786-1794 Fax: EM: sduzor@hcfa.gov
Consultant / SSA	Kestenbaum, Bert Office of the Chief Actuary Social Security Administration	Ph: (410) 965-3034 Fax: (410) 966-2658 EM: bert.m.kestbaum@ssa.gov