ASSISTED FERTILITY PROGRAM

DONATION APPLICATION FORM

FOR OFFICE USE ONLY

Donor Number _____ Revision _____

UNIFORM DONOR APPLICATION FORM

To become a sperm or egg donor, we need to learn some information about your personal and medical history. Your responses to these questions will help us to make sure that your health and medical history are compatible with the donation process and in particular for egg donors that it will not involve any increased risks for you. This effort will also help us to match you to an appropriate recipient.

Please provide complete and accurate information to these questions. If you do not know the answer, ask a parent or family member. Any information you provide during the donation process, will remain completely confidential. Some of the information from this questionnaire will be given to the recipient(s) as noted but all identifying information is removed.

A "yes" response will not necessarily eliminate you as a potential donor. Most people will have at least one of these conditions in themselves or a family member. The accuracy of the information you will be giving will provide information to potential families you may help to create.

INSTRUCTIONS:

1. **Please fill in all blanks completely**. Please complete all questions and write "N/A" if not applicable.
2. Please be specific. Avoid expressions such as "natural" or "old age" (for causes of death). List any health problems as specifically as possible. If you do not know the age, put the approximate age or ask a relative to help you. List exact relationships such as "first cousin through my mother's sister".
3. Please provide information on all the relatives requested. Do not write their names.
4. If you have any questions, please call your donor coordinator.

Last name: _____ First name: _____ Middle Initial: _____

Sex ○ Male ○ Female Age: _____

Date of Birth _____ Place of Birth _____

Soc. Security #: _____ Are you a US citizen or permanent resident? ○ Yes ○ No

Driver's License # _____ State _____

Marital Status: ○ single married ○ divorced ○ widowed ○ engaged ○ partnered

Length of Current Relationship: _____ years

_____ Date filled out _____

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DEMOGRAPHICS

MAILING ADDRESS:

Street _____ City _____

State/Province _____ Zip/ Postal code _____ Country _____

OK to leave message?

Home Phone Number _____ ○ Yes ○ No

Work Phone Number _____ ○ Yes ○ No

Cell Phone Number _____ ○ Yes ○ No

Email Address _____

Do you have medical insurance? ○ Yes ○ No

If yes, name of carrier: _____ ID # _____ Group # _____

Employer: _____

DONATION HISTORY:

Have you applied or been screened to be an egg or sperm donor before? ○ Yes ○ No

If yes, list name and location of donor program (s): _____

_____

Have you donated before? ○ Yes ○ No If yes, how many times did you donate or cycle? _____

Are you currently enrolled as an egg or sperm donor in another program? ○ Yes ○ No

---

How did you hear about our program?

☐ Radio (which station) _____ ☐ Friend (name) _____

☐ Newspaper (which one) _____ ☐ Magazine (which one) _____

☐ Website (which one) _____ ☐ Other (specify) _____

Did you consult with your family when completing your family medical history? ○ Yes ○ N**I hereby attest that all information disclosed in this application is accurate, true, and up-to-date to the best of my**

knowledge._____

(Signature of Applicant)

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PERSONAL HEALTH HISTORY

Are you currently under a physicians care for any reason? ○ Yes ○ No

If yes, please explain: _____

Have you ever had any major illnesses such as amoebic dysentery (infection of the intestine), hypertension, blood clots, pneumonia, mononucleosis, etc.? ○ Yes ○ No

If yes, when? _____

Have you had any serious illness in the past? ○ Yes ○ No

If yes, please describe: _____

Did you have any complications or concerns with anesthesia? _____

Have you had any hospitalization(s) not mentioned above? _____

_____

Please list any surgical procedures:

Have you ever had any broken bones? ○ Yes ○ No If yes, please list: _____

How many days in the preceding 12 months did you miss work because of illness (colds, flu, accidents, surgery, etc.)?

Please explain _____

Has anyone in your family, including yourself, experienced recurring and/or chronic physical symptoms that have not been evaluated by a physician (Please include those symptoms that you may not consider serious.)? ○ Yes ○ No

If yes, please describe _____

Have you ever been seen by psychiatrist, psychologist, social worker, counselor, or any other mental health professional for any reason? ○ Yes ○ No

If yes, when, for how long and for what reason? _____

Have you ever used medications such as antianxiety or antidepressants to treat an emotional or psychological problem?

○ Yes ○ No

If yes, list why and date last used _____

Have you been vaccinated in the last 6 months? ○ Yes ○ No

If yes, what were you vaccinated for? _____

List all medications that you have taken in the proceeding 12 months (prescription):

Medication	How Often	Reason

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PERSONAL HEALTH HISTORY (CONTINUED)

List all current over-the-counter medications (include hormones, vitamins, aspirin, antacids, laxatives, herbal & sports supplements, performance-enhancing supplements including steroids, etc.)

Medication	How Often	Reason

Have you ever taken anti-malarial drugs or had malaria? ○ Yes ○ No

Have you had a blood transfusion? ○ Yes ○ No If yes, when? _____

Have you ever been refused or denied as a blood donor? ○ Yes ○ No If yes, why? _____

Are you eligible to work in the United States? ○ Yes ○ No Is your work schedule flexible? ○ Yes ○ No

List all the jobs you held in the past five years:

Jobs/Duties	Year Began	Year End

Have you had radiation exposure or x-ray exposure? ○ Yes ○ No

If yes, please explain: _____

Have you ever been exposed to "agent orange" or any other herbicides or chemicals (military, forestry, highway service, or elsewhere)? ○ Yes ○ No

If yes, which substance(s)? _____

When? _____ Where? _____

In the preceding six months, were you exposed to the following in your job, living environment or while involved in hobbies? If yes to any of these, give dates and how often you have been exposed. Please consider carefully.

Exposed to	Response	When?	How Often?
Toxic Chemicals or Substances	○ Yes ○ No		
Sprays	○ Yes ○ No		
Fumes/Exhaust	○ Yes ○ No		
Radiation	○ Yes ○ No		
Flea Powder/Sprays	○ Yes ○ No		
Lead/Lead products	○ Yes ○ No		
Asbestos/Asbestos products	○ Yes ○ No		
Pesticides/Herbicides	○ Yes ○ No		
Cleaning solutions/solvents	○ Yes ○ No		

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PERSONAL HEALTH HISTORY (CONTINUED)

Do you take hot baths, saunas, hot tubs, or steam baths? ☐ Daily ☐ Weekly ☐ Occasionally ☐ Never

Within the past 6 months have you been exposed to UV rays in a tanning booth? ○ Yes ○ No

What is your caffeine usage? Number cups of coffee: _____ Soda _____ Tea _____ Energy Drinks _____

Do you currently smoke cigarettes? ☐ Daily ☐ Occasionally ☐ Rarely ☐ Never If yes, how many per day? _____

Have you ever smoked cigarettes? ○ Yes ○ No If yes, how many cigarettes per day? _____

If no, what year/month did you stop? _____

How many years did you smoke? _____

What best describes your alcohol consumption? ☐ Never drink ☐ Rarely drink/Drink in small amounts ☐ Even amounts through the week ☐ Drink in concentrated periods

What type of alcohol do you usually consume? ☐ Beer ☐ Wine ☐ Liquor

If you do drink, how many drinks do you usually consume in a week? ○ 1-3 ○ 4-9 ○ 10-15 ○ 16 or more

Have you ever used recreational or illicit drugs (cocaine, marijuana, LSD, heroin, barbiturates, narcotics, opiates, amphetamines, hallucinogens, tranquilizers, PCP, steroids, or etc.)? ○ Yes ○ No

If yes, which one (s) and when did you last use them? _____

Do you sleep well? ○ Yes ○ No If no, how do you manage this? _____

Have you had acupuncture, ear and/or body piercing or tattooing in which sterile procedures may not have been used? ○ Yes ○ No

Please list and describe all of your tattoos and body piercings:

Date Received	Description	Location on Body	Sterile Needles Used

Have you ever had any problems with the law (i.e. DUI, custody issues, lawsuits)? ○ Yes ○ No

If yes, which one (s) and when did you last use them? _____

Please list any arrests, convictions, sentences, etc. _____

Have you ever been incarcerated? ○ Yes ○ No If yes, please describe: _____

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SEXUAL AND CONTRACEPTIVE HISTORY

Sexual Orientation ☐ Homosexual ☐ Heterosexual ☐ Bisexual

Number of current sexual partners: _____ Number of sexual partners during the last six months _____

Total number of past sexual partners _____

In the last 6 months have you had unprotected sex (intercourse without a condom) with a new partner? ○ Yes ○ No

Have you ever injected drugs or had a sexual partner who did so? ○ Yes ○ No

CONTRACEPTIVE HISTORY:

Currently use: ☐ IUD Type ☐ Diaphragm ☐ Condom ☐ Birth Control Pills

☐ Rhythm ☐ Spermicide ☐ Depo-Provera ☐ Tubal Ligation ☐ None

If Birth Control Pills: _____ (name) How long on Birth Control Pills? _____

Why did you start taking Birth Control Pills? _____

If Depo-Provera, when was your last injection? _____

To your knowledge, have you or any of your sexual partners been in contact with anyone or have you been personally tested or been treated for any of the following:

	Self	Partner	If yes, when:	How many times?	When was the last time?
HIV (AIDS)					
NSU (non specific urethritis)					
Syphilis					
Gonorrhea					
Chlamydia					
Trichomonas					
Venereal Warts					
Herpes, Genital					
Viral Hepatitis B or C					
Genital Sores					
Penis Discharge					
Other sexually transmissible diseases					

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SEXUAL AND CONTRACEPTIVE HISTORY

Age at onset of menses: _____ Date of Last Menstrual Period: _____

Are your menstrual periods regular: ○ Yes ○ No

How long is your monthly cycle (first day of one period to first day of the next)? _____ days

Are you periods regular when you are not on any type of hormonal birth control such as the pill, etc.? ○ Yes ○ No

If no, how many times per year do you menstruate? _____

How many days does your period usually last? _____ days

Do you bleed or spot between periods? ○ Yes ○ No

Do you get menstrual cramps before, during, or after your period? ○ Yes ○ No

If yes, are your cramps: ○ mild ○ moderate ○ severe?

If yes, do you use medication alleviate the pain? ○ Yes ○ No

If yes, what medications do you use? _____

Have you ever had any medical treatment for menstrual problems? _____

Date of last Pap Smear: _____ Result: _____

Have you ever had an abnormal PAP ○ Yes ○ No If yes, when & why: _____

Have you ever been told you were infertile ○ Yes ○ No If yes, when & why: _____

Have you ever had a pelvic infection requiring treatment with antibiotics ○ Yes ○ No

Do you want children in the future? ○ Yes ○ No

REPRODUCTIVE HISTORY (OR PARTNER FOR SPERM DONORS)

FERTILITY HISTORY:

Number of pregnancies: _____

Number of miscarriages: _____ Date(s) of miscarriages: _____

Number of ectopic pregnancies: _____ Date(s) of ectopic pregnancy: _____

Number of abortions: _____ Date(s) of abortions: _____

Number of stillbirths: _____ Date(s) of each stillbirth: _____

Number of children: _____ Are you Currently Breastfeeding? ○ Yes ○ No

Length of time it took you or your partner to get pregnant. Shortest _____ Longest _____

Pregnancy # Boy/Girl	**Delivery Date**	**Type of Delivery** (Vaginal or C-Section)	**Complications**	**Weeks pregnant when delivered** (prematurity)	**Height / Weight**
1					
2					
3					
4					

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PLEASE NOTE THAT THE REMAINING PORTION OF THIS APPLICATION WILL BE SHARED AND VIEWED BY RECIPIENTS.

PHYSICAL CHARACTERISTICS

Are you adopted? ○ Yes ○ No Blood Type if known _____ Height _____ Weight _____

Recent weight loss/gain? ○ Yes ○ No If yes_____lbs ○ loss ○gain

What was your weight at age 21? _____

PLEASE SELECT RESPONSES THAT BEST DESCRIBE YOU BELOW:

☐ Right Handed ☐ Left Handed ☐ Ambidextrous

Bone Structure: ☐ Small ☐ Medium ☐ Large ☐ Very Large

Complexion: ☐ Very Fair ☐ Fair ☐ Light ☐ Medium ☐ Olive ☐ Light Brown ☐ Dark Brown ☐ Ebony

Tan ability: ☐ None ☐ Slight ☐ Medium ☐ Easy ☐ Freckle

Skin Condition : ☐ Oily ☐ Medium ☐ Dry ☐ Combination Dimples? ○ Yes ○ No

Eye Color: ☐ Blue ☐ Brown ☐ Lt. Brown ☐ Dark Brown ☐ Green ☐ Hazel

Eye set: ☐ Narrow ☐ Average ☐ Wide

Eye Size: ☐ Small ☐ Average ☐ Large

Shape: ☐ Round ☐ Oval ☐ Almond

Natural Hair Color: ☐ Black ☐ Light Blonde ☐ Medium Blonde ☐ Dark Blonde ☐ Light Brown ☐ Medium Brown ☐ Dark Brown ☐ Red

Hair Type: ☐ Curly ☐ Wavy ☐ Straight

Hair Texture: ☐ Fine ☐ Medium ☐ Coarse

Fullness: ☐ Thin ☐ Medium ☐ Thick

Baldness: ○ Yes ○ No **Baldness in Family** ○ Yes ○ No

Premature Graying: ○ Yes ○ No If yes, at what age _____

Body and Facial Features: ☐ Small ☐ Medium ☐ Large

Condition of your teeth: ☐ Poor ☐ Fair ☐ Good ☐ Excellent

Have you had any periodontal or orthodontic work? ○ Yes ○ No If yes, at what age?

Hearing (without corrective aids): ☐ Poor ☐ Fair ☐ Good ☐ Excellent

Vision (without corrective lenses): ☐ Poor ☐ Fair ☐ Good ☐ Excellent

Prescription (If known) _____

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PERSONAL HEALTH HISTORY
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Do you wear glasses or contacts or have you had laser surgery? ○ Yes ○ No

If yes, are/were you: ☐ Nearsighted ☐ Farsighted ☐ Other (specify): _____

Do you have astigmatism (blurred vision due to an irregularity in the curvature of the cornea.? ○ Yes ○ No

If yes, age diagnosed _____

Do you have any Allergies? ○ Yes ○ No

If yes, are they to: ☐ Food(s) ☐ Medication(s) ☐ Environmental ☐ Latex

Please list any childhood allergies that you have outgrown: _____

For each medication allergy, describe specific substance and reaction(s) and age first noticed:

Substance: _____ Reaction(s): _____ Age: _____

Substance: _____ Reaction(s): _____ Age: _____

Substance: _____ Reaction(s): _____ Age: _____

SOCIAL HISTORY AND HABITS
THIS PAGE WILL BE SHARED AND VIEWED BY RECIPIENTS

Religion Born Into: _____ Religion Practiced: _____

Grade Point Average (GPA): _____ SAT Scores: Verbal _____ Math _____ ACT Score: _____

Education:

☐ Did not Complete High School

☐ Received GED

☐ Completed high school

☐ Currently in college, pursuing degree in _____

☐ Completed college, degree in _____ GPA: _____

☐ Currently pursuing an advanced degree in _____

☐ Completed advanced degree in _____

Did you have any learning disabilities or weaknesses in school? If yes, describe: _____

Academic Strengths (i.e. math, reading): _____

How many languages do you speak? _____ Which one (s): _____

Musical Talent or Instrument: _____ Years Experience _____

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SOCIAL HISTORY AND HABITS (CONTINUED)
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Artistic Talent: _____

Athletic Skills / Favorite Sports: _____

Other skills/hobbies/talents/interests do you have (i.e. writing, reading, ability to do games or crossword puzzles, handcrafts)? Describe: _____

Current Occupation: _____ How long have you been at your current job? _____

HABITS:

Exercise Habits: ☐ None ☐ Occasional ☐ Regular ☐ Type of Exercise:

Your diet is: ☐ Vegetarian ☐ Non-vegetarian

Your diet is: ☐ Poor ☐ Average ☐ Excellent

Do you have any dietary restrictions? _____

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REPRODUCTIVE HISTORY

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YOUR CHILDREN	1	2	3	4
Age				
Sex				
Eye color				
Hair Color				
Frame size				
Grade in school				
Personality				
Artistic ability				
Intelligence				
Distinguishing characteristics				
Wears eye glasses				
Discipline problems				
Any medication				
Dyslexia				
Reading difficulties				
Speech difficulties				
Any special services at school				
Seen by Social worker/ psychiatrist				
Grade functional level: Normal / Above/ Below Average				

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FAMILY HEALTH HISTORY

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How many blood siblings are in your immediate family (including yourself and half siblings)? _____

Number of Brothers _____ Number of Sisters _____

Number of Maternal Aunts _____ Number of Maternal Uncles _____

Number of Paternal Aunts _____ Number of Paternal Uncles _____

Do you have any brothers or sisters that died in infancy or childhood? ○ Yes ○ No

If yes, what was the cause? _____

Are there any members of your family with a history of learning disabilities or autism? ○ Yes ○ No

If yes, please explain _____

Describe genetic family members according to the following characteristics. Use natural eye and hair color; fair/dark, etc. complexion. If they are deceased, please list cause of death. Please do not put "natural" as a cause of death.
If unknown, write "unknown."

	Eye Color	Hair Color	Complexion	Height	Weight	Bone Structure	Occupation/ Education	Age if living	Age at time of death	Cause of death
Sister(s)										
Brother(s)										
Mother										
Father										
Maternal Grandmother										
Materanl Grandfather										
Paternal Grandmother										
Paternal Grandfather										

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FAMILY HEALTH HISTORY (CONTINUED)
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Carefully review the following list of medical problems and identify which ones you or one of your genetic relatives have or had. Please consider each condition carefully for each family member. Explain any conditions you check below, indicating which side of the family (maternal or paternal), the age at the time of onset, and any other pertinent information. If you and none of your indicated family members have a history of the specific medical condition, please indicate none.

	None	Self	Mother	Father	Sibling	Grand parents	Aunt/ Uncle	Cousin	**Explanation** (which side of family, age of onset, etc.)
CANCER									
Breast	☐	☐	☐	☐	☐	☐	☐	☐	
Colon or Intestinal	☐	☐	☐	☐	☐	☐	☐	☐	
Lung	☐	☐	☐	☐	☐	☐	☐	☐	
Ovarian or Uterine	☐	☐	☐	☐	☐	☐	☐	☐	
Prostate or Testicular	☐	☐	☐	☐	☐	☐	☐	☐	
Skin	☐	☐	☐	☐	☐	☐	☐	☐	
Stomach	☐	☐	☐	☐	☐	☐	☐	☐	
Thyroid	☐	☐	☐	☐	☐	☐	☐	☐	
Blood (e.g. leukemia)	☐	☐	☐	☐	☐	☐	☐	☐	
Other	☐	☐	☐	☐	☐	☐	☐	☐	
HEART									
Stroke	☐	☐	☐	☐	☐	☐	☐	☐	
Heart Attack	☐	☐	☐	☐	☐	☐	☐	☐	
Congenital Heart Disease	☐	☐	☐	☐	☐	☐	☐	☐	
Heart Disease or Defect	☐	☐	☐	☐	☐	☐	☐	☐	
Hardening of the Arteries	☐	☐	☐	☐	☐	☐	☐	☐	
High Blood Pressure	☐	☐	☐	☐	☐	☐	☐	☐	
High cholesterol level	☐	☐	☐	☐	☐	☐	☐	☐	

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FAMILY HEALTH HISTORY (CONTINUED)
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	None	Self	Mother	Father	Sibling	Grand parents	Aunt/ Uncle	Cousin	**Explanation** (which side of family, age of onset, etc.)
BLOOD									
Anemia	☐	☐	☐	☐	☐	☐	☐	☐	
Sickle-Cell Anemia	☐	☐	☐	☐	☐	☐	☐	☐	
Factor V Leiden thrombpphilia (Blood clots or strokes)	☐	☐	☐	☐	☐	☐	☐	☐	
Hemophilia or other Bleeding/Clotting Disorders such as Von Willebrand's Disease	☐	☐	☐	☐	☐	☐	☐	☐	
Immune Deficiency	☐	☐	☐	☐	☐	☐	☐	☐	
Leukemia	☐	☐	☐	☐	☐	☐	☐	☐	
Lymphoma or Swollen Lymph Nodes	☐	☐	☐	☐	☐	☐	☐	☐	
HIV	☐	☐	☐	☐	☐	☐	☐	☐	
Thalassemia	☐	☐	☐	☐	☐	☐	☐	☐	
Polyarteritis Nodosa	☐	☐	☐	☐	☐	☐	☐	☐	
Other Blood Disorder	☐	☐	☐	☐	☐	☐	☐	☐	
RESPIRATORY									
Asthma	☐	☐	☐	☐	☐	☐	☐	☐	
Hay Fever	☐	☐	☐	☐	☐	☐	☐	☐	
Emphysema	☐	☐	☐	☐	☐	☐	☐	☐	
Tuberculosis	☐	☐	☐	☐	☐	☐	☐	☐	
Pneumonia	☐	☐	☐	☐	☐	☐	☐	☐	
Alpha-1 antitrypsin Disorder	☐	☐	☐	☐	☐	☐	☐	☐	
Blood in Sputum	☐	☐	☐	☐	☐	☐	☐	☐	
Other Lung Disease	☐	☐	☐	☐	☐	☐	☐	☐	

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FAMILY HEALTH HISTORY (CONTINUED)
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	None	Self	Mother	Father	Sibling	Grand parents	Aunt/ Uncle	Cousin	**Explanation** (which side of family, age of onset, etc.)
GASTRO-INTESTINAL									
Appendicitis	☐	☐	☐	☐	☐	☐	☐	☐	
Ulcer of Stomach or Duodenum	☐	☐	☐	☐	☐	☐	☐	☐	
Gallstones	☐	☐	☐	☐	☐	☐	☐	☐	
Hepatitis A,B or C	☐	☐	☐	☐	☐	☐	☐	☐	
Cirrhosis of the Liver	☐	☐	☐	☐	☐	☐	☐	☐	
Other Liver Disease	☐	☐	☐	☐	☐	☐	☐	☐	
Ulcerative Colitis	☐	☐	☐	☐	☐	☐	☐	☐	
Crohns Disease	☐	☐	☐	☐	☐	☐	☐	☐	
Pyloric Stenosis	☐	☐	☐	☐	☐	☐	☐	☐	
Multiple Polyps of the Colon	☐	☐	☐	☐	☐	☐	☐	☐	
Rectal Disorder	☐	☐	☐	☐	☐	☐	☐	☐	
Inflammatory Bowel Disease	☐	☐	☐	☐	☐	☐	☐	☐	
Any other problem of the digestive system	☐	☐	☐	☐	☐	☐	☐	☐	
METABOLIC/ ENDOCRINE									
Diabetes requiring insulin therapy	☐	☐	☐	☐	☐	☐	☐	☐	
Diabetes not requiring insulin therapy	☐	☐	☐	☐	☐	☐	☐	☐	
Childhood Diabetes	☐	☐	☐	☐	☐	☐	☐	☐	
Thyroid disorder	☐	☐	☐	☐	☐	☐	☐	☐	
Goiter	☐	☐	☐	☐	☐	☐	☐	☐	
Hypoglycemia	☐	☐	☐	☐	☐	☐	☐	☐	
Adrenal Dysfunction or Disorder	☐	☐	☐	☐	☐	☐	☐	☐	
Phenyl Ketonuria (PKU) or inherited Metabolism Disorder	☐	☐	☐	☐	☐	☐	☐	☐	
Obesity	☐	☐	☐	☐	☐	☐	☐	☐	
Dwarfism	☐	☐	☐	☐	☐	☐	☐	☐	

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FAMILY HEALTH HISTORY (CONTINUED)
THIS PAGE WILL BE SHARED AND VIEWED BY RECIPIENTS

	None	Self	Mother	Father	Sibling	Grand parents	Aunt/ Uncle	Cousin	**Explanation** (which side of family, age of onset, etc.)
URINARY									
Kidney Problems	☐	☐	☐	☐	☐	☐	☐	☐	
Polycystic Kidney Disease	☐	☐	☐	☐	☐	☐	☐	☐	
Other disease/ defect of urinary tract (urethra, bladder, ureter)	☐	☐	☐	☐	☐	☐	☐	☐	
GENITAL/ REPRODUCTIVE									
Hermaphroditism/ Ambiguous Genitals	☐	☐	☐	☐	☐	☐	☐	☐	
Hypospadias or undescended testicle	☐	☐	☐	☐	☐	☐	☐	☐	
Uterine Fibroids	☐	☐	☐	☐	☐	☐	☐	☐	
Ovarian Cysts or Ruptured	☐	☐	☐	☐	☐	☐	☐	☐	
Lumps or Cysts in Breast or Discharge	☐	☐	☐	☐	☐	☐	☐	☐	
Polycystic Ovarian Syndrome (PCOS)	☐	☐	☐	☐	☐	☐	☐	☐	
Pelvic Inflammatory Disease (PID)	☐	☐	☐	☐	☐	☐	☐	☐	
Endometriosis	☐	☐	☐	☐	☐	☐	☐	☐	
REPRODUCTIVE OUTCOMES									
2 or more Miscarriages	☐	☐	☐	☐	☐	☐	☐	☐	
Stillborn	☐	☐	☐	☐	☐	☐	☐	☐	
Premature Menopause	☐	☐	☐	☐	☐	☐	☐	☐	
Death of a newborn infant	☐	☐	☐	☐	☐	☐	☐	☐	
Childhood death	☐	☐	☐	☐	☐	☐	☐	☐	
Birth defects	☐	☐	☐	☐	☐	☐	☐	☐	
Infertility	☐	☐	☐	☐	☐	☐	☐	☐	
Premature Birth	☐	☐	☐	☐	☐	☐	☐	☐	

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FAMILY HEALTH HISTORY (CONTINUED)
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	None	Self	Mother	Father	Sibling	Grand parents	Aunt/ Uncle	Cousin	**Explanation** (which side of family, age of onset, etc.)
NEUROLOGICAL									
Migraines	☐	☐	☐	☐	☐	☐	☐	☐	
Mental retardation	☐	☐	☐	☐	☐	☐	☐	☐	
Senility or Mental Deterioration before age 50	☐	☐	☐	☐	☐	☐	☐	☐	
Multiple Sclerosis	☐	☐	☐	☐	☐	☐	☐	☐	
Cerebral Palsy	☐	☐	☐	☐	☐	☐	☐	☐	
Neurofibromatosis	☐	☐	☐	☐	☐	☐	☐	☐	
Epilepsy / Seizures	☐	☐	☐	☐	☐	☐	☐	☐	
Attention Deficit Disorder/ Hyperactivity	☐	☐	☐	☐	☐	☐	☐	☐	
Autism / Asperger's	☐	☐	☐	☐	☐	☐	☐	☐	
Alzheimer's Disease/ Dementia	☐	☐	☐	☐	☐	☐	☐	☐	
Hydrocephalus	☐	☐	☐	☐	☐	☐	☐	☐	
Tuberous Sclerosis	☐	☐	☐	☐	☐	☐	☐	☐	
Parkinson's Disease	☐	☐	☐	☐	☐	☐	☐	☐	
Creutzfeldt-Jakob Disease	☐	☐	☐	☐	☐	☐	☐	☐	
Scoliosis	☐	☐	☐	☐	☐	☐	☐	☐	
Myasthenia Gravis	☐	☐	☐	☐	☐	☐	☐	☐	
Huntington's or Wilson's Disease	☐	☐	☐	☐	☐	☐	☐	☐	
Tourette's syndrome	☐	☐	☐	☐	☐	☐	☐	☐	
Other diseases of the nervous system	☐	☐	☐	☐	☐	☐	☐	☐	

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FAMILY HEALTH HISTORY (CONTINUED)
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	None	Self	Mother	Father	Sibling	Grand parents	Aunt/ Uncle	Cousin	**Explanation** (which side of family, age of onset, etc.)
MENTAL HEALTH									
Anxiety / Panic Attacks	☐	☐	☐	☐	☐	☐	☐	☐	
Anorexia / Bulemia/ other eating disorders	☐	☐	☐	☐	☐	☐	☐	☐	
Depression	☐	☐	☐	☐	☐	☐	☐	☐	
Schizophrenia	☐	☐	☐	☐	☐	☐	☐	☐	
Manic Depressive or Bipolar Disorder	☐	☐	☐	☐	☐	☐	☐	☐	
Other mental health disorder requiring hospitalization	☐	☐	☐	☐	☐	☐	☐	☐	
Suicide Attempts	☐	☐	☐	☐	☐	☐	☐	☐	
Other mental health problems that warranted counseling (please list)	☐	☐	☐	☐	☐	☐	☐	☐	
SIGHT/SOUND/ SMELL									
Deafness before age 60	☐	☐	☐	☐	☐	☐	☐	☐	
Deformity of the ear	☐	☐	☐	☐	☐	☐	☐	☐	
Cataracts before age 50	☐	☐	☐	☐	☐	☐	☐	☐	
Blindness	☐	☐	☐	☐	☐	☐	☐	☐	
Color Blindness	☐	☐	☐	☐	☐	☐	☐	☐	
Severe Myopia	☐	☐	☐	☐	☐	☐	☐	☐	
Glaucoma	☐	☐	☐	☐	☐	☐	☐	☐	
Retinoblastoma	☐	☐	☐	☐	☐	☐	☐	☐	
Retinitis Pigmentosa	☐	☐	☐	☐	☐	☐	☐	☐	
Deviated Septum	☐	☐	☐	☐	☐	☐	☐	☐	
Any other Sensory Disorder	☐	☐	☐	☐	☐	☐	☐	☐	

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FAMILY HEALTH HISTORY (CONTINUED)
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	None	Self	Mother	Father	Sibling	Grand parents	Aunt/ Uncle	Cousin	**Explanation** (which side of family, age of onset, etc.)
MUSCLE/BONE/ JOINTS									
Muscular Dystrophy	☐	☐	☐	☐	☐	☐	☐	☐	
Achondroplasia – form of dwarfism with abnormal bone growth	☐	☐	☐	☐	☐	☐	☐	☐	
Other Chronic Muscle Disease	☐	☐	☐	☐	☐	☐	☐	☐	
Osteogenesis imperfecta (brittle bone disease)	☐	☐	☐	☐	☐	☐	☐	☐	
Loss of Muscle Coordination	☐	☐	☐	☐	☐	☐	☐	☐	
Osteoporosis	☐	☐	☐	☐	☐	☐	☐	☐	
Marfan Syndrome	☐	☐	☐	☐	☐	☐	☐	☐	
Arthritis	☐	☐	☐	☐	☐	☐	☐	☐	
Rheumatoid or Juvenile Arthritis	☐	☐	☐	☐	☐	☐	☐	☐	
Spinal Muscular Atrophy	☐	☐	☐	☐	☐	☐	☐	☐	
Hereditary Low Back Disorder or Deformity of Spine	☐	☐	☐	☐	☐	☐	☐	☐	
Reiter's Disease	☐	☐	☐	☐	☐	☐	☐	☐	
Myasthenia Gravis	☐	☐	☐	☐	☐	☐	☐	☐	
Gout	☐	☐	☐	☐	☐	☐	☐	☐	

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FAMILY HEALTH HISTORY (CONTINUED)
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	None	Self	Mother	Father	Sibling	Grand parents	Aunt/ Uncle	Cousin	**Explanation** (which side of family, age of onset, etc.)
SKIN									
Acne	☐	☐	☐	☐	☐	☐	☐	☐	
Albinism	☐	☐	☐	☐	☐	☐	☐	☐	
Eczema	☐	☐	☐	☐	☐	☐	☐	☐	
Excessive Facial Hair (Hirsutism)	☐	☐	☐	☐	☐	☐	☐	☐	
Pigmentation Disorders	☐	☐	☐	☐	☐	☐	☐	☐	
Psoriasis	☐	☐	☐	☐	☐	☐	☐	☐	
Neurofibromatosis	☐	☐	☐	☐	☐	☐	☐	☐	
Other disorders of the skin	☐	☐	☐	☐	☐	☐	☐	☐	
Infectious Skin Disease	☐	☐	☐	☐	☐	☐	☐	☐	
More than 5 purple- or coffee-colored spots on skin (size of quarter or larger)	☐	☐	☐	☐	☐	☐	☐	☐	
CHROMOSOMAL ABNORMALITIES									
Down Syndrome	☐	☐	☐	☐	☐	☐	☐	☐	
Other (i.e. Turner, Fragile X, Klinefelter's etc.)	☐	☐	☐	☐	☐	☐	☐	☐	

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FAMILY HEALTH HISTORY (CONTINUED)
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	None	Self	Mother	Father	Sibling	Grand parents	Aunt/ Uncle	Cousin	**Explanation** (which side of family, age of onset, etc.)
CONGENITAL ABNORMALITIES/ BIRTH DEFECTS									
Cleft Lip / Palate	☐	☐	☐	☐	☐	☐	☐	☐	
Congenital Hip Problems	☐	☐	☐	☐	☐	☐	☐	☐	
Club Feet	☐	☐	☐	☐	☐	☐	☐	☐	
Heart Defect	☐	☐	☐	☐	☐	☐	☐	☐	
Hearing Problems	☐	☐	☐	☐	☐	☐	☐	☐	
Spina Bifida -Neural Tube (open spine)	☐	☐	☐	☐	☐	☐	☐	☐	
Microcephaly	☐	☐	☐	☐	☐	☐	☐	☐	
Holoprosencehpaly – a single-lobed brain structure and severe skull and facial defects	☐	☐	☐	☐	☐	☐	☐	☐	
Other	☐	☐	☐	☐	☐	☐	☐	☐	
OTHER									
Alcoholism	☐	☐	☐	☐	☐	☐	☐	☐	
Drug abuse, Misuse or Addiction	☐	☐	☐	☐	☐	☐	☐	☐	
Premature degeneration of any organ system	☐	☐	☐	☐	☐	☐	☐	☐	
Any other condition not mentioned above	☐	☐	☐	☐	☐	☐	☐	☐	

More information about the above medical conditions are located at: http://www.mazornet.com/genetics/index.htm

Explain:

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GENETIC HISTORY

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ETHNIC ORIGIN (E.G., FRENCH, IRISH)

Mother: _____ Father: _____

RACE: CHECK ALL THAT APPLY FOR YOUR ANCESTORS:

African American	☐ Mother	☐ Father	☐ MGM	☐ MGF	☐ PGM	☐ PGF
Eastern European (Ashkenazi) Jewish	☐ Mother	☐ Father	☐ MGM	☐ MGF	☐ PGM	☐ PGF
Mediterranean (Greek, Italian)	☐ Mother	☐ Father	☐ MGM	☐ MGF	☐ PGM	☐ PGF
Hispanic	☐ Mother	☐ Father	☐ MGM	☐ MGF	☐ PGM	☐ PGF
Indian (from India)	☐ Mother	☐ Father	☐ MGM	☐ MGF	☐ PGM	☐ PGF
Southeast Asian (Laotian, Vietnamese, Cambodian)	☐ Mother	☐ Father	☐ MGM	☐ MGF	☐ PGM	☐ PGF
French Canadian	☐ Mother	☐ Father	☐ MGM	☐ MGF	☐ PGM	☐ PGF
Cajun	☐ Mother	☐ Father	☐ MGM	☐ MGF	☐ PGM	☐ PGF

(**MGM**=Maternal Grandmother, **MGF**=Maternal Grandfather; **PGM**=Paternal Grandmother, **PGF**=Paternal Grandfather)

Have you or anyone in your family ever been tested positive as a carrier or had any of any of the following diseases?

Blooms Syndrome	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Canavan	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Cystic Fibrosis	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Fabry Disease	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Familial Dysautonomia	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Familial Mediterranean Fever	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Fanconi Anemia Grp. C:	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Gaucher	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Niemann-Pick type A	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Mucolipidosis type IV	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Sickle Cell	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Tay-Sachs	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown
Thalassemia	○ Yes	○ No	If yes:	☐ disease	☐ carrier	☐ negative	☐ unknown

Is there anything else we should know about your family?

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PERSONAL AND MOTIVATIONAL

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In your own words, describe your personality, temperament, and character:

What physical, artistic, intellectual or social abilities do you feel best about:

What are your present and future career goals:

What are your present and future personal goals:

List the 3 achievements you are most proud of:

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PERSONAL AND MOTIVATIONAL (CONTINUED)
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What is your favorite movie? _____

What is your favorite book? _____

What is your favorite color? _____

What is your favorite food? _____

What is one of your most memorable moments and why?

If you could change one thing about yourself, what would it be and why?

Is there a person alive or dead whom you admire and why?

What would you do on a "perfect" day if you could do anything you wanted?

Describe your personality and temperament as a child:

What was your favorite thing to do as a child?

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PERSONAL AND MOTIVATIONAL (CONTINUED)
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What did your parents teach you to value?

How were you in comparison to other children?

Describe your personality and temperament as a teenager:

Did you have any problems as a child and/ or as a teenager? Explain:

Who was the most important influence on you and why?

What were your ambitions/ goals as a teenager?

What were your best and worst subjects in school?

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PERSONAL AND MOTIVATIONAL (CONTINUED)
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Please provide the following information about your family:

	Intellectual/Academic Achievements	Artistic Achievements
Mother		
Father		
Sisters		
Brothers		

Reasons for wanting to donate eggs or sperm :

If you could pass on a message to the recipient(s) of your eggs or sperm, what would that message be?

If you could write a message to the child born through your participation as an egg or sperm donor for when he/she turns 18 years old, what would you tell him/her?

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PLEASE ATTACH SEVERAL PHOTOGRAPHS OF YOURSELF

(AGES 8 – 1 YEARS, NO ADULT PHOTOS PLEASE).

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GLOSSARY- INHERITED DISEASES

DEFINITIONS

Inherited – A disease or characteristic that is transmitted through genes from parents to offspring. Inheritance patterns include the following:

Autosomal Dominant – Disorders caused by one mutated copy of a gene. An affected person usually has one affected parent. Autosomal dominant disorders usually occur in every generation of an affected family. When a person carries an autosomal dominant gene mutation, each of his/her offspring has a 50% chance for inheriting the gene mutation.

Autosomal Recessive – Disorders caused by two mutated copies of a gene. An affected person usually has unaffected parents who each carry one copy of the mutated gene. Autosomal recessive disorders are not usually seen in every generation of a family. Carrier parents have a 25% chance for having an affected child.

X-linked dominant – Disorders caused by mutations in genes located on the X chromosome. Females are more frequently affected than males, and the chance to pass on an X-linked dominant disorder differs between men and women. Fathers cannot pass the X-linked traits or disorders to their sons. Females who have an X-linked dominant gene mutation have a 50% chance to have an affected child.

X-linked recessive – Disorders caused by mutations on genes on the X chromosomes. Males are more often affected than females, and the chance to pass on the disorder differs between men and women. Families with X-linked recessive disorders often have affected males, but rarely affected females, in each generation. Females who carry an X-linked recessive gene mutation have a 50% chance to pass it on to each of her children.

Multifactorial – Disorders caused by a combination of the effects of multiple genes or by interactions between genes and the environment.

SOURCES AND ADDITIONAL INFORMATION:

Talking Glossary of Genetic Terms http://www.genome.gov/10002096; http://www.genome.gov/glossary.cfm#g

Fact Sheets http://www.genome.gov/10000202

Cancer Dictionary http://www.cancer.gov/dictionary/

Genetics Home Reference National Library of Medicine http://ghr.nlm.nih.gov/

National Institutes of Health Genetic and Rare Diseases Information Center http://rarediseases.info.nih.gov/GARD/Default.aspx?PageID=4

Gene Tests http://www.genetests.org/

GLOSSARY- INHERITED DISEASES (CONTINUED)

CANCER

- **Hereditary Breast/Ovarian Cancer** – Mutations in BRCA1 or BRCA2 genes predispose to breast cancer and ovarian cancer as well as prostate cancer (BRCA1) and other cancers (BRCA2). Hereditary breast/ovarian cancer is inherited in families in an autosomal dominant pattern. Each child of an individual with a BRCA1 or BRCA2 cancer-predisposing mutation has a 50% chance of inheriting the mutation.
- **Hereditary colon cancer**
  - **Hereditary non-polyposis colorectal cancer** - Hereditary non-polyposis colon cancer (HNPCC) is caused by an autosomal dominant inherited gene mutation. HNPCC is characterized by an increased risk of colon cancer and other cancers (e.g., of the endometrium, ovary, stomach, small intestine, hepatobiliary tract, upper urinary tract, brain, skin). Each child of an individual with a HNPCC cancer-predisposing mutation has a 50% chance of inheriting the mutation.

HEART

- **Congenital heart disease** - Congenital heart disease is a common type of birth defect or malformation in one or more structures of the heart or blood vessels that occurs during pregnancy while the fetus is developing. The cause of congenital heart disease is not known in most affected people. There are some recognized factors that are associated with an increased risk for congenital heart disease including: 1) genetic or chromosomal abnormalities such as Down syndrome; 2) taking certain medications, alcohol or drug abuse during pregnancy; and 3) maternal viral infections such as German measles in the first trimester of pregnancy. The risk of having a child with congenital heart disease is higher if a parent or a sibling has a congenital heart defect.

BLOOD

- **Sickle cell anemia** - Sickle cell disease is a group of disorders that affects hemoglobin, the molecule in red blood cells that delivers oxygen to cells throughout the body. Individuals who have sickle cell disease have atypical hemoglobin molecules called hemoglobin S, which can distort red blood cells into a sickle, or crescent, shape. Signs and symptoms include a low number of red blood cells (anemia), repeated infections, and periodic episodes of pain. The severity of symptoms varies from person to person. Sickle cell anemia is inherited in an autosomal recessive manner. Each child of carrier parents has a 25% chance to be born with sickle cell anemia.
- **Factor V Leiden thrombophilia** - Factor V Leiden thrombophilia is an inherited disorder of blood clotting. Factor V Leiden is the name of a specific mutation that results in thrombophilia - the increased tendency to form abnormal blood clots in blood vessels. People who have the factor V Leiden mutation are at somewhat higher than average risk for a type of clot that forms in veins, such as the deep veins of the legs (deep venous thrombosis), or a clot that travels through the bloodstream and lodges in the lungs (pulmonary embolism). Factor V Leiden thrombophilia can be inherited in families in an autosomal dominant and autosomal recessive manner.
- **Hemophilia** - Hemophilia is a bleeding disorder that slows the blood clotting process. People who have hemophilia often experience prolonged bleeding or oozing following an injury, surgery, or having a tooth pulled. The major types of this condition are hemophilia A (also known as classic hemophilia) and hemophilia B (also known as Christmas disease). Hemophilia A and hemophilia B are inherited in an X-linked recessive manner. In X-linked recessive inheritance, a female with one altered copy of the gene in each cell is called a carrier. She can pass on the altered gene to her children, but usually does not experience signs and symptoms of the disorder
- **Tay-Sachs** - Tay-Sachs disease is a rare inherited disorder that causes progressive destruction of nerve cells in central nervous system (the brain and spinal cord). Affected infants progressively lose motor skills such as turning over, sitting, and crawling. Children who have the severe infantile form of Tay-Sachs disease usually survive only into early childhood. Tay-Sachs disease is inherited in an autosomal recessive manner. Carrier parents have a 25% in each pregnancy to have an affected child.
- **Thalassemia** - Beta thalassemia is an inherited blood disorder that reduces the production of hemoglobin. Symptoms of beta thalassemia occur when not enough oxygen gets to various parts of the body due to low levels of hemoglobin and a shortage of red blood cells. Beta thalassemia is inherited in an autosomal recessive manner. Carrier parents have a 25% chance in each pregnancy to have an affected child.

GLOSSARY- INHERITED DISEASES (CONTINUED)

RESPIRATORY

- **Alpha-1 antitrypsin disorder** - Alpha-1 antitrypsin deficiency is an inherited condition that can cause lung disease in adults and liver disease in adults and children. This disorder is inherited in an autosomal co-dominant pattern. Co-dominance means that two different versions of the gene may be expressed, and both versions contribute to the genetic trait.

GASTROINTESTINAL

- **Cystic Fibrosis** - Cystic fibrosis is an inherited disorder of the mucus glands that affects many body systems. The most common signs and symptoms of cystic fibrosis include progressive damage to the respiratory system and chronic digestive system problems. Cystic fibrosis is inherited in an autosomal recessive manner. Carrier parents have a 25% chance in each pregnancy for having an affected child.
- **Pyloric stenosis** - Pyloric stenosis (also called infantile pyloric stenosis or gastric outlet obstruction) is a condition that involves a narrowing of the pylorus, the lower part of the stomach through which food and other stomach contents pass to enter the small intestine. When an infant has pyloric stenosis, the muscles in the pylorus become enlarged to the point where food is prevented from emptying out of the stomach. Pyloric stenosis is known to run in families. When a parent has pyloric stenosis, then, their infant has an increased risk of developing the disorder.

METABOLIC/ENDOCRINE

- **Phenylketonuria** - Phenylketonuria (also known as PKU) is an inherited disorder that increases the levels of a substance called phenylalanine in the blood. Phenylalanine is a building block of proteins that is obtained through the diet. If PKU is not treated, phenylalanine can build up to harmful levels in the body, causing mental retardation and other serious health problems. PKU is inherited in an autosomal recessive manner. Carrier parents have a 25% chance with each pregnancy to have an affected child.
- **Dwarfism** – There are a number of different types of dwarfism and many are inherited in families. Examples of types of dwarfism include: achondroplasia, thanatophoric dysplasia, and Robinow syndrome.

URINARY

- **Polycystic kidney disease** - Polycystic kidney disease is a disorder that affects the kidneys and other organs. Cysts, develop in the kidneys, causing them to become enlarged and can lead to kidney failure. Cysts may also develop in other organs, particularly the liver. There are two major forms of polycystic kidney disease distinguished by the age of onset and their pattern of inheritance. The autosomal dominant form (sometimes called ADPKD) has signs and symptoms that typically begin in adulthood, although cysts in the kidney are often present from childhood. The autosomal recessive form of polycystic kidney disease (sometimes called ARPKD) is much rarer and is often lethal early in life.

GENITAL/REPRODUCTIVE

- **Hypospadias** – Hypospadias is a birth defect of the urethra that happens in males. It involves an abnormally placed opening in the penis. Instead of opening at the tip of the penis, a hypospadic urethra opens anywhere along the line running from the tip along the underside of the shaft to the where the penis and scrotum meet. In most males hypospadias is not inherited, nor is their family recurrence. In some cases, hypospadias happens as a result of a chromosomal abnormality called a pericentric inversion of chromosome number 16.

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GLOSSARY- INHERITED DISEASES (CONTINUED)

REPRODUCTIVE OUTCOMES

- **2 or more miscarriages** – Miscarriage (also called spontaneous abortion) is the term used for a pregnancy that ends on it's own, within the first 20 weeks of gestation. The causes of miscarriages are varied, and most often the cause cannot be identified. During the first trimester, the most common cause of miscarriage is chromosomal abnormality - meaning that something is not correct with the baby's chromosomes. In some cases the chromosome abnormality in the developing fetus is the result of a parent carrying a balanced chromosomal arrangement called a translocation. This can lead to multiple miscarriages.
- **Birth defects** – A birth defect is a problem that happens while the baby is developing in the mother's body. Most birth defects happen during the first 3 months of pregnancy. A birth defect can affect almost any part of the body. Causes of birth defects include a family history of birth defects, maternal age, certain drugs taken during pregnancy, alcohol use and smoking during pregnancy.

NEUROLOGICAL

- **Mental Retardation** - Mental retardation is a term used to describe a person who has certain limitations in mental functioning and difficulties in communicating, taking care of him or herself, and social skills. These limitations will cause a child to learn and develop more slowly than a typical child. Causes of mental retardation include genetic conditions such as Down syndrome, problems during pregnancy, problems at birth and health problems such as malnutrition.
- **Cerebral palsy** - Cerebral palsy is the term for a group of disorders that involve the loss of movement or loss of other nerve function. Cerebral palsy is caused by injuries to the largest part of the brain (cerebrum) which happen as the baby grows in the womb or near the time of birth. There are multiple causes of cerebral palsy including birth defects that affect the brain, spinal cord, head, face, lungs or metabolism, and certain hereditary and genetic conditions.
- **Neurofibromatosis** – There are two types of neurofibromatosis. Neurofibromatosis type 1 is a disorder characterized by changes in skin coloring (pigmentation) and the growth of tumors along nerves in the skin, brain, and other parts of the body. The signs and symptoms of this condition vary widely among affected people. Neurofibromatosis type 1 is considered to have an autosomal dominant pattern of inheritance. Neurofibromatosis type 2 is a disorder characterized by the growth of noncancerous tumors in the nervous system. Neurofibromatosis type 2 is also considered to have an autosomal dominant pattern of inheritance. However, unlike most other autosomal dominant conditions, in which one altered copy of a gene in each cell is sufficient to cause the disorder, two copies of the NF2 gene must be altered to trigger tumor formation in neurofibromatosis type 2. A mutation in the second copy of the NF2 gene happens in other cells in the nervous system during a person's lifetime. Almost everyone who is born with one NF2 mutation acquires a second mutation in many cells and develops the tumors characteristic of neurofibromatosis type 2
- **Autism/Aspergers** –
  - Autism and autism spectrum disorders are complex neurodevelopmental conditions. The genetics of autism are complex and it is thought that there are multiple genes involved.
  - Aspergers – Asperger syndrome is one of several autism spectrum disorders, with symptoms of difficulty in social interactions and restricted, stereotyped interests and activities. Children who have Aspergers syndrome do not usually have language or cognitive developmental delays. Genes are believed to play a role in Aspergers syndrome, and it seems to run in some families.
- **Hydrocephalus** – Hydrocephalus is a condition in which the primary characteristic is excessive accumulation of fluid in the brain. The excessive accumulation of fluid causes an abnormal widening of spaces in the brain called ventricles. This widening creates potentially harmful pressure on the tissues of the brain. The causes of hydrocephalus are still not well understood. Hydrocephalus may be caused by inherited genetic abnormalities (such as the genetic defect that causes aqueductal stenosis) or developmental disorders (such as those associated with neural tube defects including spina bifida and encephalocele). Other possible causes include complications of premature birth, and diseases such as tumors or hemorrhage which block the fluid..

GLOSSARY- INHERITED DISEASES (CONTINUED)

- **Tuberous sclerosis** - Tuberous sclerosis is a genetic disorder characterized by the growth of numerous noncancerous tumors in many parts of the body. These tumors can occur in the skin, brain, kidneys, and other organs, in some cases leading to significant medical problems. Tuberous sclerosis is inherited in an autosomal dominant manner, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In about one-third of families, an affected person inherits an altered gene from a parent who has the disorder. About two thirds of cases result from new gene mutations. These cases occur in people with no history of tuberous sclerosis in their family.
- **Creutzfeldt-Jakob Disease** – Creutzfeldt-Jakob disease is a prion disease. Prion diseases are group of progressive conditions that affect the nervous system. Prion diseases impair brain function, causing memory changes, personality changes, a decline in intellectual function, and problems with movement that worsen over time. The signs and symptoms of these conditions usually begin in adulthood, and these disorders lead to death within a few months to several years. Only a small percentage of prion disease cases run in families. Most cases occur in people without any known risk factors or gene mutations. Creutzfeldt-Jakob disease is acquired by eating beef products obtained from cattle that have prion disease.
- **Huntington Disease** - Huntington disease is a progressive brain disorder that causes uncontrolled movements, mental and emotional problems, and loss of thinking ability. Adult-onset Huntington disease, is the most common form of this disorder, with onset usually in a person's thirties or forties. An early-onset, less common form of Huntington disease begins in childhood or adolescence. This condition is inherited in an autosomal dominant manner, which means one copy of the altered gene in each cell is sufficient to cause the disorder.
- **Gaucher Disease** - Gaucher disease is an inherited disorder that affects many of the body's organs and tissues. The signs and symptoms of this condition vary widely among affected individuals. There are several types of Gaucher disease based on their particular features. Some types do not affect the brain and spinal cord while others do. Type 1 Gaucher disease, for example, is the most common form of this disorder. Major signs and symptoms of Type 1 Gaucher disease include enlargement of the liver and spleen, a low number of red blood cells, easy bruising caused by a decrease in blood platelets, lung disease, and bone abnormalities such as bone pain, fractures, and arthritis. Types 2 and 3 Gaucher disease, on the other hand, have problems that affect the central nervous system. Gaucher disease is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents each carry one copy of the mutated gene, but they do not show signs or symptoms of the disease.
- **Wilson's Disease** - Wilson disease is an inherited disorder in which excessive amounts of copper accumulate in the body, particularly in the liver, brain, and eyes. Typically, signs and symptoms of Wilson disease first appear during the teenage years. Wilson's disease is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents each carry one copy of the mutated gene, but they do not show signs or symptoms of the disease.
- **Tourette syndrome** - Tourette syndrome is a complex disorder characterized by repetitive, sudden, and involuntary movements or noises called tics. Tics usually appear in childhood, and their severity varies over time. In most cases, tics become milder and less frequent in late adolescence and adulthood. Individuals who have Tourette syndrome are also at risk for other associated problems including attention deficit hyperactivity disorder (ADHD), obsessive-compulsive disorder (OCD), anxiety, depression, and problems with sleep. A variety of genetic and environmental factors appear to play a role in causing Tourette syndrome. Most of these factors are unknown to date. Among family members of an affected person, it is therefore difficult to predict who else may be at risk of developing the condition.

MENTAL HEALTH

- **Depression** – Clinical depression is an illness that can challenge a person's ability to perform even routine daily activities, and in some cases lead a person to contemplate or commit suicide. There are several different types of depression (mood disorders that include depressive symptoms) such as major depression, bipolar disorder and seasonal depression. The causes of depression are complex. Genetic, biological, and environmental factors can contribute to its development. In some people, depression can be traced to a single cause, while in others, a number of causes are involved. For many, the causes are never known. Certain types of depression seem to run in some families. Research is ongoing as to exactly which genes are involved in depression.

GLOSSARY- INHERITED DISEASES (CONTINUED)

MUSCLE/BONE JOINT

- **Muscular dystrophy** - Muscular dystrophies are a group of genetic conditions characterized by progressive muscle weakness and wasting. The Duchenne and Becker types of muscular dystrophy primarily affect the skeletal muscles, which are used for movement, and the muscles of the heart. These conditions occur much more frequently in males than in females. Both Duchenne and Becker muscular dystrophy are inherited in an X-linked recessive pattern, with the mutated gene that causes the disorder on the X chromosome. Males are affected by X-linked recessive disorders much more frequently than females.
- **Achondroplasia** - Achondroplasia is a disorder of bone growth. particularly in the long bones of the arms and legs. All people with achondroplasia have short stature. Health problems commonly associated with achondroplasia include breathing difficulties (called apnea), obesity, and recurrent ear infections. Achondroplasia is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. About 80 percent of individuals with achondroplasia have average-size parents; these cases result from a new gene mutation in that individual. In the remaining cases, people with achondroplasia have inherited a gene from one or two affected parents.
- **Osteogenesis imperfecta** - Osteogenesis imperfecta (OI) is a group of genetic disorders that mainly affect the bones. People who have OI have bones that break easily, often from mild trauma or with no apparent cause. Multiple fractures are common, and in severe cases, fractures can occur even before birth. Milder cases may involve only a few fractures over a person's lifetime. There are at least eight recognized forms of osteogenesis imperfecta, designated type I through type VIII, distinguished by their signs and symptoms. Most types of osteogenesis imperfecta have an autosomal dominant pattern of inheritance, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Many people with type I or type IV osteogenesis imperfecta inherit a mutation from a parent who has the disorder.
- **Marfan syndrome** - Marfan syndrome is a connective tissue disorder. Connective tissue provides strength and flexibility to structures throughout the body such as bones, ligaments, muscles, the walls of blood vessels, and heart valves. Marfan syndrome affects most organs and tissues, especially the skeleton, lungs, eyes, heart, and the large blood vessel that distributes blood from the heart to the rest of the body called the aorta. Individuals who have Marfan syndrome often are tall and slender, have elongated fingers and toes, a long narrow face, highly arched palate, and have an arm span that exceeds their body height. About half of all people with Marfan syndrome have vision problems caused by a dislocated lens (ectopia lentis) Most people with Marfan syndrome have abnormalities of the heart and the aorta. This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is needed to cause the disorder. At least one quarter of classic Marfan syndrome cases result from a new gene mutation. These individuals have no history of the disorder in their family.
- **Spinal muscular atrophy** - Spinal muscular atrophy is a disorder that affects the control of muscle movement. It is caused by a loss of specialized nerve cells, (motor neurons), in the spinal cord and the part of the brain that is connected to the spinal cord (the brainstem). The loss of motor neurons leads to weakness and shrinkage of muscles used for activities such as crawling, walking, sitting up, and controlling head movement. In severe cases of spinal muscular atrophy, the muscles used for breathing and swallowing are affected. There are a number of different subtypes of spinal muscular atrophy based on the age of onset and symptoms. Most types of spinal muscular atrophy are inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. One type of spinal muscular atrophy is inherited in an autosomal dominant manner, which means one copy of the altered gene in each cell is sufficient to cause the disorder.
- **Reiter's disease** - Reiter's syndrome, also known as reactive arthritis, is a type of arthritis that occurs as a reaction to an infection somewhere in the body. Most infections that cause the disease begin in the bladder, urethra, penis, or vagina and are spread through sexual intercourse, a form of the disease called genitourinary Reiter's syndrome, or urogenital Reiter's syndrome. Other infections that can cause reactive arthritis include gastrointestinal infections due to eating contaminated food or handling contaminated substances, a form of the disease called gastrointestinal Reiter's syndrome, or enteric Reiter's syndrome. Reiter's syndrome affects mostly young men, between the ages of 20 and 40. Although researchers are not sure why some people develop reactive arthritis in response to certain infections, a genetic factor (presence of the HLA-B27 gene) appears to increase the risk.

GLOSSARY- INHERITED DISEASES (CONTINUED)

SIGHT/SOUND/SMELL

- **Deafness** – There are several types of deafness including conductive hearing loss, neural hearing loss (nerve deafness), and mixed hearing loss (a combination of conductive and neural hearing loss). Some people are born deaf. Usually the cause is unknown. Although deafness is inherited in some families, deaf parents often have hearing children and hearing parents often have deaf children. Diseases and injuries of the ear can also cause deafness.
- **Blindness** – Blindness is a condition of lacking visual perception that is due to physiological or neurological factors. Blindness has a number of causes including disease and malnutrition. Blindness may have a genetic cause, and may also be a symptom of a particular genetic disorder. Recent advances in mapping of the human genome have identified genetic causes of low vision or blindness, for example the disorder called Bardet-Biedl syndrome.
- **Color blindness** – Color blindness is the inability to perceive differences between some of the colors that other people can distinguish. It is usually genetic in nature, but may also be due to eye, nerve or brain damage, or to exposure to certain chemicals. Color blindness can be inherited in families. Since the mapping of the human genome there have been many causative gene mutations discovered. Mutations capable of causing color blindness originate from at least 19 different chromosomes and many different genes.
- **Retinoblastoma** - Retinoblastoma is a rare type of eye cancer that develops in the retina, the part of the eye that detects light and color. Although this disorder can occur at any age, it usually develops in young children. Most cases of retinoblastoma occur in only one eye, but both eyes can be affected. Retinoblastoma can be inherited in an autosomal dominant pattern which means that one copy of the altered gene in each cell is sufficient to increase cancer risk. A person with retinoblastoma may inherit an altered copy of the gene from one parent, or the altered gene may be the result of a new mutation. For retinoblastoma to develop, a second mutation in the other copy of the RB1 gene must occur in retinal cells during the person's lifetime. When there is a family history of retinoblastoma or if the person develops tumors in both eyes, the gene mutation is probably in all of the person's cells, and that person is said to have an inherited form of retinoblastoma. A smaller number of individuals have retinoblastoma as a result of missing portions of chromosome 13 that are not inherited.

SKIN

- **Albinism** – Albinisim is a condition in which there is a lack of melanin pigment in the eyes, skin and hair (or more rarely the eyes alone). Albinisim is hereditary and results from inheritance of recessive gene mutations. There are two main categories of Albinism - 1) oculocutaneous albinism in which there is a lack of melanin pigment in skin and hair, and 2) ocular albinism, in which only the eyes lack pigment. People with oculocutaneous albinism can have anywhere from no pigment at all to almost-normal levels. People who have ocular albinism have generally normal skin and hair color, and many even have a normal eye appearance. Albinism may also be a feature of a genetic syndrome such as Hermansky-Pudlak syndrome.
- **Neurofibromatosis** – There are two types of Neurofibromatosis – Type 1 and Type 2. Neurofibromatosis type 1 is a disorder characterized by changes in skin coloring and the growth of tumors along nerves in the skin, brain, and other parts of the body. The signs and symptoms of this condition vary widely among affected people. Neurofibromatosis type 2 is a disorder characterized by the growth of noncancerous tumors in the nervous system. The most common develop along the nerve that carries information from the inner ear to the brain (the auditory nerve). Tumors that occur on nerves in other areas of the brain or spinal cord are also commonly seen with this condition. Both Type 1 and Type 2 Neurofibromatosis are considered to have an autosomal dominant pattern of inheritance. People with Neurofibromatosis Type 1 and Type 2 are born with one mutated copy of either the NF1 or NF2 mutated genes in each cell. In about half of cases, the gene mutation is inherited from an affected parent. The remaining cases result from new mutations in the gene and occur in people with no history of the disorder in their family. Unlike most other autosomal dominant conditions, in which one altered copy of a gene in each cell is sufficient to cause the disorder, two copies of either the NF1 or NF2 gene must be altered to trigger tumor formation in neurofibromatosis. A mutation in the second copy of the NF1 or NF2 gene occurs during a person's lifetime in specialized cells surrounding nerves. Almost everyone who is born with one NF1 or NF2 mutation acquires a second mutation in many cells and develops the tumors characteristic of the disease.

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GLOSSARY- INHERITED DISEASES (CONTINUED)

CONGENITAL ABNORMALITIES/BIRTH DEFECTS

- **Cleft lip/palate** – Cleft lip and palate are common birth defects that affect the upper lip and the roof of the mouth. There are many causes of cleft lip and palate. Gene alterations passed down from one or both parents, drugs used or maternal viruses during pregnancy can cause cleft lip and/or palate. Cleft lip and palate can also be part of a genetic syndrome or occur with other birth defects. Risk factors for cleft lip and palate also include a family history of cleft lip or palate and other birth defects.
- **Congenital hip problems** – Congenital hip problems, also called hip dysplasia, involve problems with formation of the hip joint in children. The location of the hip dysplasia can be either the ball of the hip joint (femoral head), the socket of the hip joint (the acetabulum), or both. Hip dysplasia, called congenital dysplasia of the hip (or CDH) in the past is now called developmental dysplasia of the hip (DDH). There are a number of factors that contribute to cause DDH. One known risk factor is having a family history of hip dysplasia. Other causes include when the baby is born in breech position or when there is a lack of intrauterine fluid (oligohydramnios) during pregnancy.
- Club feet – Clubfoot is a condition where the foot turns inward and downward. It is a congenital condition, meaning it is present at birth. Other terms for clubfoot are Talipes equinovarus and Talipes. Clubfoot is the most common congenital disorder involving the legs, and can range from mild and flexible to severe and rigid. Although the exact cause is not known, clubfoot may be passed down in some families. Family history, therefore, is a risk factor for clubfoot, as is being a male.
- **Heart Defect** - A congenital heart defect involves an abnormal structure of the heart that is present at birth. Congenital heart defects are the most common type of major birth defect. There are multiple causes of congenital heart defects including environmental and genetic factors. Genes that can cause congenital heart defects are now being discovered, such as a gene that can cause an atrial septal defect and one that may contribute to hypoplastic left heart syndrome. Congenital heart defects can also be a part of a wider pattern of birth defects and genetic syndromes such as Down syndrome, Turner syndrome and velocardiofacial syndrome.
- **Hearing problems** - There are several types of hearing loss including conductive hearing loss, neural hearing loss (nerve deafness), and mixed hearing loss (a combination of conductive and neural hearing loss). Some people are born with hearing loss. Usually the cause is unknown. Although hearing loss is inherited in some families, deaf parents often have hearing children and hearing parents often have deaf children. Diseases and injuries of the ear can also cause deafness.
- **Spina bifida** - Spina bifida is one of a group of birth defects called neural tube defects. Spina bifida occurs during fetal development when a portion of the neural tube fails to develop or close properly causing defects in the spinal cord and in the bones of the backbone. Spina bifida, like many other birth defects appears to be caused by a combination of genetic and environmental risk factors, such as a family history of neural tube defects, folic acid deficiency, and medical conditions such as diabetes and obesity.
- **Microcephaly** - Microcephaly is disorder in which the circumference of the head is smaller than normal because the brain has not developed properly or has stopped growing. Microcephaly can be present at birth or it may develop in the first few years of life. It is most often caused by genetic abnormalities that interfere with the growth of the cerebral cortex during the early months of fetal development. Microcephaly is associated with genetic syndromes such as Down syndrome, chromosomal syndromes, and neurometabolic syndromes, Babies may also be born with microcephaly if their mother abuses drugs or alcohol during pregnancy, or becomes infected with the German measles, chicken pox.
- **Holoprosencephaly** - Holoprosencephaly is a disorder caused by the failure of the embryonic forebrain (prosencephalon) to divide properly into the double lobes of the cerebral hemispheres. As a result, the baby has a single-lobed brain structure and severe skull and facial defects. In most cases of holoprosencephaly, the malformations are so severe that babies die before birth. In less severe cases, babies are born with normal or near-normal brain development and facial deformities that may affect the eyes, nose, and upper lip. Often, no specific cause for holoprosencephaly can be identified. There are some specific chromosomal abnormalities that have been identified as the cause of holoprosencephaly in some patients. In some families, holoprosencephaly is inherited in autosomal dominant or X-linked recessive inheritance. Several genes have also been identified that play a role in causing holoprosencephaly.

GLOSSARY- INHERITED DISEASES (CONTINUED)

CHROMOSOMAL ABNORMALITIES

- **Down syndrome** - Down syndrome is a chromosomal disorder that is associated with mental retardation, a characteristic facial appearance, and poor muscle tone in infancy. Individuals who have Down syndrome may also have heart defects, digestive problems such as gastroesophageal reflux or celiac disease, hearing loss, and cancer of blood-forming tissue (leukemia). Some people with Down syndrome have hypothyroidism. Down syndrome also appears to be is associated with an increased risk of Alzheimer disease Down syndrome is usually caused by the presence of an extra chromosome number 21, called trisomy 21, which means each cell in the body has three copies of chromosome 21 instead of the usual two copies. Most cases of Down syndrome are not inherited, but occur as random events during the formation of egg or sperm. One type of Down syndrome, called translocation Down syndrome, can be inherited.
- **Fragile X syndrome** - Fragile X syndrome is a genetic disorder that involves a range of developmental problems including learning disabilities and mental retardation, and behavioral problems such as hyperactive behavior and attention deficit disorder. Males are usually more severely affected by this disorder than females. Many males with fragile X syndrome have characteristic physical features that become more apparent with age such as a long and narrow face, large ears, prominent jaw and forehead, unusually flexible fingers, and enlarged testicles after puberty. Most cases of fragile X syndrome are caused by a mutation in which a DNA segment, known as the CGG triplet repeat, is expanded within the FMR1 gene. Fragile X syndrome is inherited in families in an X-linked dominant pattern.
- **Turner syndrome** - Turner syndrome is a chromosomal disorder that affects development in females. Women with Turner syndrome are often shorter than average and are usually unable to conceive children because they lack ovarian function. Other features of Turner syndrome can include extra skin on the neck, puffiness or swelling of the hands and feet, skeletal abnormalities, heart defects, and kidney problems. Developmental delays, learning disabilities, and behavioral problems are may also be present, although these characteristics vary among affected females. In most cases, Turner syndrome is not inherited. Rather, it occurs as random events during the formation of egg or sperm.
- **Klinefelter syndrome** - Klinefelter syndrome is a chromosomal disorder that affects male sexual development. Most males who have Klinefelter syndrome have one extra copy of the X chromosome in each cell. The presence of an extra X chromosome interferes with male sexual development causing their testicles to develop abnormally, and leading to low levels of the hormone testosterone beginning during puberty. A lack of testosterone can lead to breast development, reduced facial and body hair, and an inability to father children. Boys who have Klinefelter syndrome may have learning disabilities and difficulty with speech and language development. Klinefelter syndrome is caused by the presence of one or more extra copies of the X chromosome in a male's cells. Klinefelter syndrome is not inherited, but usually occurs as a random event during the formation of egg or sperm.

GENETIC HISTORY

- **Bloom syndrome** - Bloom syndrome is an inherited disorder that is characterized by a high frequency of breaks and rearrangements in an affected person's chromosomes. Individuals who have Bloom syndrome are usually much smaller than average, and often have a high-pitched voice and characteristic facial features including a long, narrow face; small lower jaw; and prominent nose and ears. They tend to develop pigmentation changes that often appear as a butterfly-shaped patch of reddened skin on the face. Other features of the Bloom syndrome may include learning disabilities, mental retardation, chronic lung problems, diabetes, and immune deficiency that leads to recurrent pneumonia and ear infections. Men with Bloom syndrome are usually not able to father children because they do not produce sperm. Women with the disorder generally experience menopause earlier than usual. Chromosome instability in Bloom syndrome also results in a high risk of cancer in affected individuals. Bloom syndrome is inherited in families in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations.

GLOSSARY- INHERITED DISEASES (CONTINUED)

- **Canavan disease** - Canavan disease is an inherited disorder that causes progressive damage to nerve cells in the brain. The signs and symptoms of Canavan disease usually begin in early infancy; however, the course of the disorder can be quite variable. Infants with Canavan disease usually appear normal for the first few months of life. By age 3 to 5 months, these infants begin to have developmental delays in motor skills, weak muscle tone, large head size, and mental retardation. They may also develop feeding and swallowing difficulties, seizures, and sleep disturbances. Canavan disease is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations.
- **Fabry Disease** - Fabry disease is an inherited disorder that begins in childhood and results from the buildup of a particular type of fat in the body's cells. Characteristic features of Fabry disease include episodes of pain, particularly in the hands and feet; clusters of small, dark red spots on the skin; a decreased ability to sweat; cloudiness of the front part of the eye; and hearing loss. Individuals with Fabry disease are also at risk for potentially life-threatening complications such as progressive kidney damage, heart attack, and stroke. Fabry disease is inherited in an X-linked pattern; however, unlike other X-linked disorders, Fabry disease causes significant medical problems in many females who have one altered copy of the mutated gene. These women may experience many of the classic features of the disorder.
- **Familial Dysautinomia** - Familial dysautonomia is a genetic disorder that affects the development and survival of certain nerve cells. The disorder causes disturbances in autonomic nerve cells, which control involuntary actions such as digestion, breathing, production of tears, and the regulation of blood pressure and body temperature. It also affects activities related to the senses, such as taste and the perception of pain, heat, and cold. Familial dysautonomia is also called hereditary sensory and autonomic neuropathy, type III. Problems related to this disorder first appear during infancy and include poor muscle tone, feeding difficulties, poor growth, lack of tears, frequent lung infections, and difficulty maintaining body temperature. Developmental delays in walking and speech, are usually present, although some affected individuals do not show signs of developmental delay. Familial dysautinomia is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations.
- **Familial Mediterranean Fever** - Familial Mediterranean fever is an inherited disorder that involves recurrent episodes of painful inflammation in the abdomen, chest, or joints. These episodes are often accompanied by fever and sometimes a rash. The first episode usually occurs by the age of 20. For some affected individuals, however, the initial episode occurs much later in life. The episodes usually last 12 to 72 hours and may vary in severity and length of time between attacks. A buildup of protein deposits occurs in some cases of familial Mediterranean fever and this can lead to kidney failure if left untreated. This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have the mutations. Rarely, familial Mediterranean fever may be inherited in an autosomal dominant pattern, which means one copy of an altered gene is sufficient to cause the disorder.
- **Fanconi Anemia** - Fanconi anemia is a rare, inherited blood disorder that causes bone marrow failure. Fanconi anemia causes the bone marrow to stop making enough new blood cells for the body to function normally. Infants born with Fanconi anemia are at higher risk for having birth defects. Fanconi anemia can also cause the bone marrow to make many abnormal blood cells, which can lead to serious health problems such as cancer. This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have the mutations.
- **Niemann-Pick, Type A** - Niemann-Pick disease is an inherited disorder that involves lipid metabolism - the breakdown, transport, and use of fats and cholesterol in the body. In affected individuals the abnormal lipid metabolism causes harmful amounts of lipids to accumulate in the spleen, liver, lungs, bone marrow, and brain. There are four main types of Niemann-Pick disease. Type A presents during infancy and is characterized by an enlarged liver and spleen, failure to thrive, and progressive deterioration of the nervous system. Children born with Niemann-Pick, Type A generally do not survive past early childhood. Niemann-Pick, Type A is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations.
- **Mucolipidosis Type IV** - Mucolipidosis Type IV is a genetic disorder, primarily which is characterized by severe neurological and ophthalmologic abnormalities. Also known as ML4, the disorder usually presents during the first year of life with mental retardation, corneal opacities, and delayed motor milestones. Children with ML4 begin to show signs of developmental delay during their first year of life. They usually attain a maximum developmental age of 15 months in language and motor function, although their receptive abilities are more advanced. They may also experience retinal degeneration that severely limits their vision. ML4 is inherited in an autosomal recessive pattern which means both copies of the gene in each cell have mutations.