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### **OOCYTE PERSONAL HISTORY FORM**

Date filled out: 02/16/2012 Donor #: 0347 (Agency Use Only) **(Incomplete applications will not be accepted)**

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To become an 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 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.
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I am interested in an ( ) Open ( ) Anonymous ( ) Semi-Open-Donation **( ) No Preference**

Are you willing to travel for an egg donation? X Yes \_\_\_\_ No Possibly if: \_\_\_\_\_

Are you currently enrolled as an egg in another program? \_\_\_\_ Yes X No

#### **How did you hear about our program?**

- |  |  |
|--|--|
| <input type="checkbox"/> Radio (which station) _____ | <input type="checkbox"/> Friend (name) _____                         |
| <input type="checkbox"/> Newspaper (which one) _____ | <input type="checkbox"/> Magazine (which one) _____                  |
| <input type="checkbox"/> Website (which one) _____   | <input checked="" type="checkbox"/> Other (specify) <u>Contacted</u> |

**\*Once you are chosen as an egg donor, first morning appointments are mandatory. You are not able to choose the dates of your appointments, as they will coincide with the clinic’s requirements and timing of your cycle. You will be required to attend 7-10 monitoring appointments prior to the actual egg retrieval. Most clinics are open between 7:30-8:00 a.m. If your schedule cannot accommodate this criterion, you will not qualify for the egg donation process.**

## DEMOGRAPHICS

Street: \_\_\_\_\_ City: Lake Jackson

State/Province: TX Zip/ Postal code: \_\_\_\_\_ Country: \_\_\_\_\_

### OK to leave message?

Home Phone Number: ( ) \_\_\_\_\_ - \_\_\_\_\_ ☒ Yes ☐ No

Work Phone Number: ( ) \_\_\_\_\_ - \_\_\_\_\_ ☒ Yes ☐ No

Cell Phone Number: ( ) \_\_\_\_\_ - \_\_\_\_\_ ☐ Yes ☐ No

Text Messaging Acceptable? ☒ Yes ☐ No

Email Address: \_\_\_\_\_ Alternate Email: \_\_\_\_\_

I check my email: ☒ all day ☐ once a day ☐ several times a week ☐ rarely

Do you have medical insurance? X Yes ☐ No

If yes, name of carrier: \_\_\_\_\_ ID #: \_\_\_\_\_ Group #: \_\_\_\_\_

Employer: \_\_\_\_\_

Who may we contact in case of an emergency? \_\_\_\_\_

Relationship \_\_\_\_\_ Ph: \_\_\_\_\_

Who may we contact in case your demographics change? \_\_\_\_\_ Ph: \_\_\_\_\_

Height: 5'7" Weight: 150 lbs **Measurements:** Bust 36 Hips \_\_\_\_\_ Waist \_\_\_\_\_

Recent weight loss/gain? ☐ Yes ☒ No If yes \_\_\_\_\_ lbs loss/gain Blood Type: O Rh Factor ☒ + or -

**Ethnic origin:** (e.g., German, Irish, English, African American, Italian, Mexican, Asian, etc...)

Mother: English, German Father: German, Irish

Last name: \_\_\_\_\_ First name: \_\_\_\_\_ Middle Initial: \_\_\_\_\_

Age: 26 Date of Birth: 07/26/1985 Place of Birth Galveston, TX

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: 3 Months/ ☒ Years

**Please circle responses that best describe you below:**

Right Handed Left Handed Ambidextrous

**Bone Structure:** Small Medium Large Very Large

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

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

**Skin Condition:** Oily Medium Dry Combination **Dimples?** X 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 Lt. Blonde Med. Blonde Dk. Blonde Lt. Brown Med. Brown

Dk. Brown Auburn Red

**Hair Type:** Curly Wavy Straight **Hair Texture:** Fine Medium Coarse **Fullness:** Thin Medium Thick

**Baldness:** \_\_\_ Yes X No **Baldness in Family:** \_\_\_ Yes X No

**Premature Graying:** \_\_\_ Yes X 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 X 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): N/A

Do you wear glasses or contacts or have you had laser surgery? \_\_\_ Yes X 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 X No

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

Please list any childhood allergies that you have outgrown: None

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

Substance: \_\_\_\_\_ Reaction(s): \_\_\_\_\_ Age: \_\_\_\_\_

Substance: \_\_\_\_\_ Reaction(s): \_\_\_\_\_ Age: \_\_\_\_\_

## MENSTRUAL AND REPRODUCTIVE HISTORY

Age at onset of menses: 12

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)? 28 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? 3 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? No

Date of last Pap Smear: 6/1/2011 Result: Normal

Have you ever had an abnormal PAP: No If yes, when & cause:

Have you ever been told you were infertile: 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

### **FERTILITY HISTORY:**

Number of pregnancies: 3

Number of miscarriages: \_\_\_\_\_

Number of ectopic pregnancies: \_\_\_\_\_

Number of abortions: \_\_\_\_\_

Number of stillbirths: \_\_\_\_\_

Number of children: 4

Length of time it took you or your partner to get pregnant. Shortest 1 day Longest 1 week

Date(s) of miscarriages: \_\_\_\_\_

Date(s) of ectopic pregnancy: \_\_\_\_\_

Date(s) of abortions: \_\_\_\_\_

Date(s) of each stillbirth: \_\_\_\_\_

Are you Currently Breastfeeding? ☐ Yes ☒ No

Did your mother take DES while she was pregnant with you? No

**Diethylstilbestrol (DES, former BAN stilboestrol)** is a synthetic nonsteroidal estrogen that was first synthesized in 1938. It is also classified as an endocrine disruptor. Human exposure to DES occurred through diverse sources, such as dietary ingestion from supplemented cattle feed and medical treatment for certain conditions, including breast and prostate cancers. From about 1940 to 1970, DES was given to pregnant women in the mistaken belief it would reduce the risk of pregnancy complications and losses. In 1971, DES was shown to cause a rare vaginal tumor in girls and women who had been exposed to this drug in utero. The United States Food and Drug Administration subsequently withdrew DES from use in pregnant women. Follow-up studies have indicated DES also has the potential to cause a variety of significant adverse medical complications during the lifetimes of those exposed.<sup>[1]</sup> The United States National Cancer Institute recommends<sup>[2]</sup> women born to mothers who took DES undergo special medical exams on a regular basis to screen for complications as a result of the drug. Individuals who were exposed to DES during their mothers' pregnancies are commonly referred to as "DES daughters" and "DES sons". Wikipedia

Pregnancy # Boy/Girl	Delivery Date	Type of Delivery (Vaginal or C- Section)	Complications	Weeks pregnant when delivered (prematurity)	Height / Weight
1 Girl	3/10/05	Vaginal	None	40	6.1 /21"
2. Girl	5/2/06	Vaginal	None	39	9.5/23"
3. Girl	5/25/11	C-section	None	36	5.7/17"
4. Girl	5/25/11	C-section	None	36	6.2/18"
5.					
6.					

YOUR CHILDREN	1	2	3	4
Age	6	5	8 months	8 months
Sex	F	F	F	F
Eye color	Blue	Blue	Blue	Blue
Hair Color	Lt. brown	Blonde	Brown	Auburn
Frame size	Small	Med	Small	Med
Grade in school	1st	K	N/A	N/A
Personality	Outgoing, fun	Reserved, athletic		
Artistic ability	Sings, dances	Draws		
Intelligence	High	High	High	High
Distinguishing characteristics	Curly Hair	Eyes	Smile	Big blue eyes
Wears eye glasses	No	No	No	No
Discipline problems	No	No	No	No
Any medication	No	No	No	No
Dyslexia	No	No	No	No
Reading difficulties	No	No	No	No
Speech difficulties	No	No	No	No
Any special services at school	No	No	No	No
Seen by Social worker/ psychiatrist	No	No	No	No
Grade functional level: Normal / Above/ Below Average	Average	Above Average		

## DONATION HISTORY

Have you applied or been screened to be an egg donor before? \_\_\_\_Yes    XNo    If yes, list name and location of donor program (s):

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Have you ever donated your eggs before? Yes If yes, Please list dates and outcomes:

Mo/Year	# Eggs Retrieved	# Eggs Fertilized	Did a pregnancy occur?	Did a live birth occur?
10/2012	22	Unsure	Yes	Pending

Were their embryos left to cryopreserve (freeze)? Info Pending If yes, approximately how many per cycle? \_\_\_\_\_

How soon are you able to commit to a donation? ASAP

What is the compensation you are asking for your donation? \$7000.00 (1<sup>st</sup> time donors \$5,000)

What is the least amount you would consider? ?

Will you require missed wages from work? Negotiable

If yes, what is your hourly wage? 22/hr      How many hours per week do you work? 40

Will you require childcare reimbursement? No    If yes, what is the hourly rate? \_\_\_\_\_ X \_\_\_\_\_ kids

During travel assignments, will you:   
☒ Drive yourself to the airport and require parking reimbursement   
☐ Take a taxi or shuttle and require reimbursement   
☒ Have someone drop you off and require NO reimbursement

Will you require high speed internet access in your hotel to keep up with work or school? XYes    \_\_\_\_No

## PERSONAL HEALTH HISTORY

Are you currently under a physicians care for any reason? \_\_\_\_ Yes X 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 X No

If yes, when? \_\_\_\_\_

Have you had any serious illness in the past? \_\_\_\_ Yes X No

If yes, please describe: \_\_\_\_\_

Did you have any complications or concerns with anesthesia? No

Have you had any hospitalization(s) not mentioned above? No

Please list any surgical procedures: C-Section

Have you ever had any broken bones? \_\_\_\_ Yes X 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: Stomach virus-2 days

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 X 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 X No

If yes, when, for how long and for what reason? \_\_\_\_\_

Have you ever used medications such as anti anxiety or antidepressants to treat an emotional or psychological problem? \_\_\_\_ Yes X No

If yes, list why and date last used \_\_\_\_\_

Have you been vaccinated in the last 6 months? \_\_\_\_ Yes X No

If yes, what were you vaccinated for? \_\_\_\_\_

**List all prescription medications that you have taken in the preceding 12 months:**

Medication	How Often	Reason
None		

**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
None		

Have you ever taken anti-malarial drugs or had malaria? \_\_\_\_ Yes X No

Have you had a blood transfusion? \_\_\_\_ Yes X No If yes, when? \_\_\_\_\_

Have you ever been refused or denied as a blood donor? \_\_\_\_ Yes X No If yes, why? \_\_\_\_\_

Do you have frequent nose bleeds, bleeding gums while brushing your teeth and or clots with menstrual periods?  
No

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

Have you had excessive radiation exposure or x-ray exposure? \_\_\_\_ Yes X 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 X 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	<u>No</u>		
Sprays	Yes	<u>No</u>		
Fumes/Exhaust	Yes	<u>No</u>		
Radiation	Yes	<u>No</u>		
Flea Powder/Sprays	Yes	<u>No</u>		
Lead/Lead products	Yes	<u>No</u>		
Asbestos/Asbestos products	Yes	<u>No</u>		
Pesticides/Herbicides	Yes	<u>No</u>		
Cleaning solutions/solvents	<u>Yes</u>	No	Occasionally	Weekly, house cleaning



What is your caffeine usage? Number cups of coffee: 0 Soda 0 Tea 1 Energy Drinks 0

Do you currently smoke cigarettes? Daily Occasionally Rarely **Never** If yes, how many per day? \_\_\_\_\_

Have you ever smoked cigarettes? \_\_\_\_ Yes X 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  
X Rarely drink/Drink in small amounts \_\_\_\_ Even amounts through the week \_\_\_\_ Drink in concentrated periods

What type of alcohol do you usually consume? \_\_\_\_ Beer X Wine X Liquor

If you do drink, how many drinks do you usually consume in a week? X 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 X No  
If yes, which one (s) and when did you last use them? \_\_\_\_\_

Do you sleep well? X 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 X No

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

Date Received:	Description:	Location on Body:	Sterile Needles Used?
1990	Piercing	Ears	Yes

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

If yes, please explain \_\_\_\_\_

Please list any arrests, convictions, sentences, incarcerations, etc.:

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**\*To become an egg donor, you must be completely drug free. You must also be nicotine free for at least 6 mos. You may be tested for this substance at any time.**

## SEXUAL AND CONTRACEPTIVE HISTORY

Sexual Orientation (please circle or highlight): Heterosexual Homosexual Bisexual

Number of current sexual partners: 1 Number of sexual partners during the last six months: 1

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

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

Currently use: IUD Type \_\_\_\_\_ Diaphragm \_\_\_\_\_ Condom \_\_\_\_\_ Birth Control Pills \_\_\_\_\_

Rhythm \_\_\_\_\_ Spermicide \_\_\_\_\_ Depo-Provera \_\_\_\_\_ Tubal Ligation \_\_\_\_\_ None X

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: NONE**

	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					
Other sexually transmissible diseases					

## SOCIAL HISTORY AND HABITS

Religion Born Into: Christianity Religion Practiced Christianity

H.S. Grade Point Average (GPA): 3.7 SAT Scores: Verbal \_\_\_\_\_ Math \_\_\_\_\_ ACT Score: \_\_\_\_\_

Education: \_\_\_\_\_ Did not Complete High School  
\_\_\_\_\_ Received GED  
\_\_\_\_\_ Completed high school  
\_\_\_\_\_ Completed/enrolled in Vocational Schooling \_\_\_\_\_  
\_\_\_\_\_ Currently in college, pursuing degree in \_\_\_\_\_  
☒ Completed college, degree in Business Administration GPA: 3.4  
\_\_\_\_\_ Currently pursuing an advanced degree in \_\_\_\_\_  
\_\_\_\_\_ Completed advanced degree in \_\_\_\_\_

Please list names and year of all colleges attended:

	<u>College</u>	<u>Year</u>
1.	<u>University of Phoenix</u>	<u>2008-2010</u>
2.	_____	_____
3.	_____	_____

Dean's List or Honor Roll? \_\_\_\_\_

Did you have any learning disabilities or weaknesses in school? If yes, describe: No

Academic Strengths (i.e. math, reading): Math, Science

How many languages do you speak? 1 Which one (s): English

Musical Talent or Instrument: Singing, piano Years Experience: 26  
Artistic Talent: No

I consider myself: Athletic ☒ Active Average Inactive

Have you excelled in any physical activities? ☒ Yes \_\_\_\_\_ No If Yes, which one(s)? Soccer

Manual Dexterity: Dexterous ☒ Average Clumsy

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

Current Occupation: Executive Account Manager How long have you been at your current job? 2 years

### HABITS:

Exercise Habits: \_\_\_\_\_ None \_\_\_\_\_ Occasional ☒ Regular Type of Exercise: Cardio, jogging

Your diet is: \_\_\_\_\_ Vegetarian \_\_\_\_\_ Non-vegetarian \_\_\_\_\_ Vegan Your diet is: poor ☒ average excellent

Do you have any dietary restrictions? Limit caffeine and sugar.

## FAMILY HEALTH HISTORY

**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
Mother	Brown	Brown	Fair	5'4"	150	Avg	Homemaker/ High school	43		
Father	Hazel	Brown	Fair	6'3"	175	Avg	Carpenter/ Trade School	45		
Brother 1.	Blue	Brown	Fair	6'2"	140	Small	Student	16		
2.										
3.										
Sister 1.										
2.										
3.										
Maternal Grandmother	Green	Brown	Fair	5'3"	130	Small	Artist/H.S.	65		
Maternal Grandfather	Blue	Brown	Fair	5'8"	140	Avg	Electrician/ Bachelors	67		
Paternal Grandmother	Blue	Red	Fair	5'7"	140	Avg	Accountant/ Masters	70		
Paternal Grandfather	Hazel	Brown	Med	6'3"	200	Avg	Carpenter/ Trade School	71		

Did you consult with your family when completing your family medical history? ☒ Yes ☐ No

Are you adopted? ☐ Yes ☒ No If Yes, are you listing your biological history? ☐ Yes ☐ No

If Yes, do you have access to your biological history? ☐ Yes ☐ No

## GENETIC HISTORY

**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___

Do you have any brothers or sisters that died in infancy or childhood? \_\_\_Yes \_\_\_XNo

If yes, what was the cause? \_\_\_\_\_

Are there any members of your family with a history of learning disabilities or autism? \_\_\_Yes \_\_\_XNo

If yes, please explain \_\_\_\_\_

(**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	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Canavan	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Cystic Fibrosis (Caucasian)	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Fabry Disease	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Familial Dysautonomia	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Familial Mediterranean Fever	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Fanconi Anemia Grp. C:	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Fragile X	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Gaucher	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Niemann-Pick type A	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Mucopolidosis type IV	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Sickle Cell (African American)	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Spinal Muscular Atrophy	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Tay-Sachs (Jewish)	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___
Thalassemia (Greek/Italian)	<b>No</b>	If yes: ___disease___	___carrier___	___negative___	___unknown___

Have you had a chromosomal analysis performed? No If so, were your results of normal XX karyotype? \_\_\_\_\_

Is there anything else we should know about your family?

No

## PERSONAL AND MOTIVATIONAL

**In your own words, describe your personality, temperament, and character:** I am very hard working, easy going and motivated person. I love to be involved with my work, my children's activities and love to be in a leadership roles. I am a huge planner and always have everything scheduled.

**What physical, artistic, intellectual or social abilities do you feel best about:** I love to socialize and meet new people. I am in many groups that allow me to practice public speaking and come into contact with new people every day scheduled.

**What are your present and future career goals :**

Currently I am in insurance. I work as an operations manager for an agency. In the future, I would like to do insurance seminars and insurance training.

**What are your present and future personal goals:** We are remodeling our home to sell and find our dream home. I can't wait to move and decorate our new home. I would love to vacation more as well.

**List the 3 achievements you are most proud of:**

My beautiful daughters

That I came from a broken home and became the well rounded, independent person I am

Recently I was interviewed by Fox news and featured on a consumer report

**What is your favorite movie?** My Fair Lady

**What is your favorite book?** Hunger Games

**What is your favorite color?** Pink

**What is your favorite food?** Italian

**What is one of your most memorable moments and why?** Going to Disney World in high school. It was an amazing trip and the friend that I went with passed away the next year, it made it that much more special.

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

I tend to over-comment and get stressed out in the process. I wish I could say "no" better and take on less.

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

My mother. She has accomplished a lot in her life despite not ever having much.

**What would you do on a “perfect” day if you could do anything you wanted?**

Sleep! and spend an uninterrupted day with my family-completely stress free. That would be perfect ☺

**Describe your personality and temperament as a child:**

I was high spirited, loved making new friends and trying new things.

**What was your favorite thing to do as a child?**

I loved to play pretend. I would be a teacher, lawyer, waitress; anything that I could create lesson plans or schedules for at my desk. I loved paperwork and writing.

**What did your parents teach you to value?**

I was taught to value myself and to never take anything for granted.

**How were you in comparison to other children?**

Maybe a little more buy but all around, I was normal. I loved being in plays and singing-anything to be the center of attention.

**Describe your personality and temperament as a teenager:**

I was very mature for my age and had a take charge attitude. I was involved in a lot and was a leader in most of the clubs/activities I was in.

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

Other than dealing with having a stepdad and the obstacles that brings, not really.

**Who was the most important influence on you and why?**

My mom, because we have always been best friends.

**What were your ambitions/ goals as a teenager?**

I wanted to be a special education teacher, a lawyer, a singer-maybe all! I had very high goals and of course they were always changing.

**What were your best and worst subjects in school?**

Favorite was math and even though I did good in it, I disliked history.

**Please provide the following information about your family:**

	Intellectual/Academic Achievements	Artistic Achievements
Mother		Paints
Father		Beautiful carpentry work
Sisters		
Brothers	4.0 GPA in H.S.	Plays Guitar

**Reasons for wanting to donate eggs:** I love to help others and I cannot imagine the devastation of being infertile. I would love to help a couple out that deserves a baby, parenthood has been one of the best gifts I've been given.

**Do you consider yourself a reliable person?** Yes

**Do you consider yourself a punctual person?** Yes

**Would you describe yourself as a religious or spiritual person?** Yes

**Do you have any ethical, moral or religious reservations about being an egg donor?** No

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

I would love to be picked by a couple that will make amazing parents-becoming a parent is such an amazing experience and I am excited to help a family become whole.

**Would you be willing to donate to gay or single prospective parents?** Yes **Please specify:** Either

**Would you be willing to meet a child conceived as the result of your donation?** Yes **Please elaborate:**

If the family and child were okay with it, absolutely but I don't have to.

**Would you be interested in possibly meeting the prospective parents or are you OK with them knowing your first name?**

Yes, this is a very intimate experience and I would like the parents to be comfortable and will be accepting to whatever they choose.

**What are your beliefs concerning selective reduction or aborting a fetus due to an anomaly(s) (birth defect)?**

I personally would have reservations if I were carrying, however, it is not my life and would respect the decision of the parents.



**How do you feel about the possibility of any remaining embryos being donated to another infertile couple that cannot afford the cost of infertility treatment? Will you sign a consent permitting such donation?**

Possibly as long as it wasn't to more than one or two other couples.

**How do you feel about any remaining embryos being discarded or donated to scientific research? Will you sign a consent permitting such medical or scientific research or destruction of such remaining embryos?**

Yes

**Some clinics have their Prospective Parents sign away rights to any remaining embryos to be used at the clinic's discretion, how do you feel about not knowing the outcome of their decision?**

Yes

**What are your thoughts on the possibility of this couple using a gestational carrier (surrogate) or a qualified sperm donor?**

Whatever gets the job done!

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

I donated my eggs to bless a family with you and I pray that your life is wonderful and fulfilling. Everyone has a purpose in life and I hope you have found or will find yours. I know that you have brought endless joy to your family's life!

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.

**\*PLEASE REFER TO THE GLOSSARY ON THE LAST PAGES OF THIS FORM FOR DEFINITIONS**

	<u>None</u>	<u>Self</u>	<u>Mother</u>	<u>Father</u>	<u>Sibling</u>	<u>Grand- parents</u>	<u>Aunt/ Uncle</u>	<u>Cousin</u>	<u>Explanation (which side of family, age of onset, etc.)</u>
<b><u>CANCER</u></b>									
Breast	X								
Colon or Intestinal	X								
Lung	X								
Ovarian or Uterine	X								
Prostate or Testicular	X								
Skin	X								
Stomach	X								
Thyroid	X								
Blood (e.g. leukemia)	X								
Other	X								
<b><u>HEART</u></b>	X								
Stroke	X								
Heart Attack	X								
Congenital Heart Disease	X								
Heart Disease or Defect	X								
1. from birth	X								
2. lifestyle	X								
High cholesterol level							X		M-aunt-Diet

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

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

<b><u>URINARY</u></b>									
Kidney Problems	X								
Polycystic Kidney Disease	X								
Other disease/ defect of urinary tract (urethra, bladder, ureter)	X								
<b><u>GENITAL/ REPRODUCTIVE</u></b>	X								
Hermaphroditism/ Ambiguous Genitals	X								
Hypospadias or undescended testicle	X								
Uterine Fibroids	X								
Ovarian Cysts or Ruptured	X								
Lumps or Cysts in Breast or Discharge	X								
Polycystic Ovarian Syndrome (PCOS)	X								
Pelvic Inflammatory Disease (PID)	X								
Endometriosis	X								
<b><u>REPRODUCTIVE OUTCOMES</u></b>									
2 or more Miscarriages	X								
Stillborn	X								
Premature Menopause	X								
Death of a newborn infant	X								
Childhood death	X								
Birth defects	X								
Infertility	X								
Premature Birth	X								

	<u>None</u>	<u>Self</u>	<u>Mother</u>	<u>Father</u>	<u>Sibling</u>	<u>Grand- parents</u>	<u>Aunt/ Uncle</u>	<u>Cousin</u>	<u>Explanation (which side of family, age of onset, etc.)</u>
<b><u>NEUROLOGICAL</u></b>									
Migraines	X								
Mental retardation	X								
Senility or Mental Deterioration before age 50	X								
Multiple Sclerosis	X								
Cerebral Palsy	X								
Neurofibromatosis	X								
Epilepsy / Seizures	X								
Attention Deficit Disorder/ Hyperactivity	X								
Autism / Asperger's	X								
Alzheimer's Disease/Dementia	X								
Hydrocephalus	X								
Tuberous Sclerosis	X								
Parkinson's Disease	X								
Creutzfeldt-Jakob Disease	X								
Scoliosis	X								
Myasthenia Gravis	X								
Huntington's or Wilson's Disease	X								
Tourette's syndrome	X								
Other diseases of the nervous system	X								
<b><u>MENTAL HEALTH</u></b>									
Anxiety / Panic Attacks	X								
Anorexia / Bulimia/other eating disorders	X								

	<u>None</u>	<u>Self</u>	<u>Mother</u>	<u>Father</u>	<u>Sibling</u>	<u>Grand- parents</u>	<u>Aunt/ Uncle</u>	<u>Cousin</u>	<u>Explanation (which side of family, age of onset, etc.)</u>
Depression	X								
Schizophrenia	X								
Manic Depressive or Bipolar Disorder	X								
Other mental health disorder requiring hospitalization	X								
Suicide Attempts	X								
Other mental health problems that warranted counseling (please list)	X								
<b><u>MUSCLE/BONE JOINTS</u></b>									
Muscular Dystrophy	X								
Achondroplasia – form of dwarfism with abnormal bone growth	X								
Other Chronic Muscle Disease	X								
Osteogenesis imperfecta (brittle bone disease)	X								
Loss of Muscle Coordination	X								
Osteoporosis	X								
Marfan Syndrome	X								
Arthritis	X								
Rheumatoid or Juvenile Arthritis	X								
Spinal Muscular Atrophy	X								
Hereditary Low Back Disorder or Deformity of Spine	X								
Reiter's Disease	X								
Myasthenia Gravis	X								
Gout	X								

	<u>None</u>	<u>Self</u>	<u>Mother</u>	<u>Father</u>	<u>Sibling</u>	<u>Grand- parents</u>	<u>Aunt/ Uncle</u>	<u>Cousin</u>	<u>Explanation (which side of family, age of onset, etc.)</u>
Metabolic Bone Disease (be more specific)	X								
Lupus (systemic lupus erythematosus – SLE)	X								
<b><u>SIGHT/SOUND/ SMELL</u></b>									
Deafness before age 60	X								
Deformity of the ear	X								
Cataracts before age 50	X								
Blindness	X								
Color Blindness	X								
Severe Myopia	X								
Glaucoma	X								
Retinoblastoma	X								
Retinitis Pigmentosa	X								
Deviated Septum	X								
Any other Sensory Disorder	X								
<b><u>SKIN</u></b>									
Acne	X								
Albinism	X								
Eczema	X								
Excessive Facial Hair (Hirsutism)	X								
Pigmentation Disorders	X								
Psoriasis	X								
Neurofibromatosis	X								



	<u>None</u>	<u>Self</u>	<u>Mother</u>	<u>Father</u>	<u>Sibling</u>	<u>Grand- parents</u>	<u>Aunt/ Uncle</u>	<u>Cousin</u>	<u>Explanation (which side of family, age of onset, etc.)</u>
Other disorders of the skin	X								
Infectious Skin Disease	X								
More than 5 purple- or coffee- colored spots on skin (size of quarter or larger)	X								
<b><u>CONGENITAL ABNORMALITIES/ BIRTH DEFECTS</u></b>									
Cleft Lip / Palate	X								
Congenital Hip Problems	X								
Club Feet	X								
Heart Defect	X								
Hearing Problems	X								
Spina Bifida -Neural Tube (open spine)	X								
Microcephaly	X								
Holoprosencephaly – a single-lobed brain structure and severe skull and facial defects	X								
Other	X								
<b><u>CHROMOSOMAL ABNORMALITIES</u></b>									
Down Syndrome	X								
Other (i.e. Turner, Fragile X, Klinefelter's etc.)	X								
<b>OTHER</b>									
Alcoholism	X								
Drug abuse, Misuse or Addiction	X								
Premature degeneration of any organ system	X								
Any other condition not mentioned above	X								

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

**CONSENT FORM**

Under the penalty of perjury, I attest that all of the information I have provided in my donor application is true and complete to the best of my knowledge. I also have a good understanding of the commitment involved in the egg donation process.

Donor's Printed Name: \_\_\_\_\_

Donor's Signature: \_\_\_\_\_

Date: \_\_\_\_\_

I \_\_\_\_\_ give An Eggceptional Match, LLC full authority to include my photographs on its web site, and to share my personal profile with prospective parents but without disclosing to them any of my identifying information. I hereby release An Eggceptional Match, LLC and Angela Bevill, and their agents, employees, successors and assigns, from any and all liability of any nature whatsoever as a result of such disclosures.

Donor's Signature: \_\_\_\_\_

Date: \_\_\_\_\_

## 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.

## 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 hypospadiac 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.

## 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.

## GLOSSARY- INHERITED DISEASES (continued)

- **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.
- **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.

## GLOSSARY- INHERITED DISEASES (continued)

- **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.

### 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.



## GLOSSARY- INHERITED DISEASES (continued)

- **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.

### 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.

## GLOSSARY- INHERITED DISEASES (continued)

- **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** – Albinism is a condition in which there is a lack of melanin pigment in the eyes, skin and hair (or more rarely the eyes alone). Albinism 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.

### 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.

## GLOSSARY- INHERITED DISEASES (continued)

- **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 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 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 Dysautonomia** - 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 dysautonomia 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-

## GLOSSARY- INHERITED DISEASES (continued)

Pick, Type A is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations.

- **Mucopolipidosis Type IV** - Mucopolipidosis 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.

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