



PO Box 1646  
Castle Rock, Colorado 80104  
(Ph) 720-733-0184 · (Fax) 720-733-2433  
**info@donatedeggs.com**  
**www.donatedeggs.com**

### **OOCYTE PERSONAL HISTORY FORM**

Date of Application: 02/02/2013 Date of Birth: 11/22/1991 Age: 21 Donor #: **0371** (Agency Use Only)

How did you hear of An Eggceptional Match? (If website, pls. specify): Friend who has been a donor

The type of donation I am interested in is (Check all that apply): ( ) Open ( ) Anonymous ☒ No Preference

---

---

***(Incomplete applications will not be accepted)***

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.

**\*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 7:00-8:00 a.m. If your schedule cannot accommodate this criterion, you will not qualify for the egg donation process.**

## DEMOGRAPHICS

Full Legal Name and any aliases: \_\_\_\_\_

Social Security #: \_\_\_\_\_ Insurance Co: \_\_\_\_\_ Farmers \_\_\_\_\_

Address: \_\_\_\_\_ City: Colorado Springs State: CO Zip: \_\_\_\_\_

Home Phone: \_\_\_\_\_ Work Phone: \_\_\_\_\_

Cell Phone: \_\_\_\_\_ May we leave a voicemail message at: (Pls. Circle): Home Work Cell

Are email communications permissible? If so, what is your E-mail Address: \_\_\_\_\_

I check my email: ( all day) once a day several times a week rarely (alternate E-mail) N/A

Are text messages permissible and if so at what telephone numbers? Yes No \_\_\_\_\_

Are you currently listed with any other clinics or agencies? No If yes, whom? N/A

Have you signed a contract with any other clinic or agency? No If so, please provide a complete copy to me.

Have you ever been denied entry into another egg donor program? No If yes, please explain in detail: N/A

How soon are you able to begin your donation? ASAP

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

Relationship: Roommate/Best friend Ph: \_\_\_\_\_

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

Marital Status: X single \_\_\_\_\_ married \_\_\_\_\_ divorced \_\_\_\_\_ widowed \_\_\_\_\_ engaged \_\_\_\_\_ partnered

Length of Current Relationship: \_\_\_\_\_ Months/ Years Are you a U.S. Citizen? (Yes) No

Do you have medical insurance? (Yes) No

If so, provide name of your health plan and identification number: Tricare Prime

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

Do you have any lawsuits or other legal claims pending against you? Yes (No)

Have you ever filed bankruptcy? Yes (No) If so, when? \_\_\_\_\_

Driver's License #: \_\_\_\_\_ State of Issue: CO

## PHYSICAL CHARACTERISTICS

Height: 5'02"    Weight: 115 lbs.    **Measurements:** Bust \_\_\_\_\_ Hips \_\_\_\_\_ Waist \_\_\_\_\_

Recent weight loss/gain? No    If Yes: lbs. \_\_\_\_\_ loss/gain

Race: White/Asian    (Caucasian, African American, Asian, Hispanic, etc...)

Ethnicity: (Pls. Be specific, ie...German, French, Irish, etc...) Korean/German

Mother's Side: Korean

Father's Side: German

Blood Type: A (+ or -)    Place of Birth: Colorado springs, CO

What celebrity do people most commonly say you look like? Brenda Song

### PLEASE CIRCLE (OR HIGHLIGHT) APPROPRIATE RESPONSE

<b>Body Type/Bone Structure:</b>		<u>(small)</u>	medium	large	very large
<b>Hands:</b>		<u>(right-handed)</u>	left-handed	ambidextrous	
<b>Eyes:</b>	*Color	brown	<u>(hazel)</u>	green	blue    gray
	*Set	narrow	average	<u>(wide)</u>	
	*Size	small	average	<u>(large)</u>	
	*Shape	round	oval	<u>(almond)</u>	
	*Shade	light	<u>(medium)</u>	dark	
<b>Hair:</b>	*Natural Color	blond	<u>(brown)</u>	black	red    other _____
	*Color as child	blond	<u>(brown)</u>	black	red
	*Shade	light	medium	<u>(dark)</u>	
	*Type	straight	<u>(wavy)</u>	curly	
	*Fullness	thin	medium	<u>(thick)</u>	
	*Texture	<u>(fine)</u>	medium	course	
<b>Nose:</b>	*Size	<u>(small)</u>	medium	large	
	*Width	narrow	<u>(average wide)</u>		
	*length	short	<u>(average wide)</u>		
	*Nostril Flare	<u>(small)</u>	average wide		
<b>Cheekbones:</b>	*Set	low	<u>(average)</u>	high	
	*Prominence	slight	<u>(medium)</u>	strong	
<b>Mouth:</b>	*Size	small	average	<u>(large)</u>	
	*Lips	thin	average	<u>(full)</u>	

<b>Chin:</b>	*Shape	square	oval	(round)			
	*Prominence	(slight)	average strong				
	*Cleft	(none)	slight	medium			
<b>Skin:</b>	*Tone	light	(med-light)	medium	med-dark	dark	olive
	*Tan Ability	none	slight	medium	(easy)		
	*Condition	(normal)	dry	oily	medium	combination	
	*Acne	(none)	slight	medium	severe	at what age	_____
<b>Other Facial Features:</b>	*Moles	(none)	one	several	numerous		
	*Freckles	(none)	several	moderate	numerous		
	*Dimples	(none)	slight	medium	deep		
<b>Eyesight:</b>	*Vision	normal	(far-sighted)	near-sighted			
	*Glasses	none	(single)	bifocal			
	*Astigmatism	yes	(no)	age diagnosed	_____		
<b>Hearing:</b> (Without corrective aids):	Poor	Fair	Good	(Excellent)			
<b>Dental:</b>	*Device	none	(braces)	retainer other	_____		
	*Reason	(cosmetic)	accident	disease	other	_____	
	*Age during use	8 to 15	years of age				

### REPRODUCTIVE/CONTRACEPTIVE HISTORY

Age at onset of menses? 13 Are your cycle's regular? Yes Date of Last Menstrual Period: Jan 17, 2013

Are your periods regular when you are not on any type of hormonal birth control such as the pill, etc.? Yes

If no: How many times per year do you menstruate? N/A

Have you ever had any medical treatment for menstrual problems? No

How long are your cycles from day one to the next day one? 28-30 days How many days do they last? 7

Do you spot or bleed between periods? No

Do you experience cramps? None (Mild) Average Severe

Have you been sexually active in the past 6 months? Yes

Are you currently sexually active? No If yes, is it a monogamous relationship? N/A If yes, for how long? N/A

If no, will your partner consent to standard blood testing? N/A

Have you or your partner ever had a sexually transmitted disease (trichomonias, chlamydia, syphilis, condyloma, gonorrhea, herpes)? Yes (No) If yes, when and what was your treatment regimen? N/A

Date of last Pap Smear: May 5, 2012

Have you ever been diagnosed with any gynecological problems such as endometriosis, ovarian cysts, abnormal Pap smears, fibroids, polyps? Yes ( **No** ) If yes, please explain treatment N/A

Currently use: IUD Type \_\_\_\_\_ Diaphragm \_\_\_\_\_ Condom \_\_\_\_\_ Birth Control Pills \_\_\_\_\_ Rhythm \_\_\_\_\_  
Spermicide \_\_\_\_\_ Nuva Ring \_\_\_\_\_ Tubal Ligation \_\_\_\_\_ Vasectomy \_\_\_\_\_ None **X**

If Birth Control Pills: N/A (name) How long on Birth Control Pills? N/A

Why did you start taking Birth Control Pills? N/A

If Depo-Provera, when was your last injection? N/A

Have you ever been pregnant? No If yes, did you have trouble conceiving? N/A

Do you want (more) children in the future? N/A

Have you ever been treated for infertility? 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

### LIST OF PREGNANCIES AND OUTCOMES

Year	Delivery ♀ or ♂ Section/Vag	Miscarriage	Ectopic	Blighted Ovum	Termination
1.					
2.					
3.					
4.					
5.					
6.					

Any complications? N/A

## **DONATION HISTORY**

Have you applied or been screened to be an egg donor before? \_\_\_\_ Yes ☒ No

If yes, list name and location of donor program (s): N/A

---

---

---

Have you ever donated your eggs before? \_\_\_\_ No \_\_\_\_ If yes, Please list dates and outcomes:

Mo/Year	# Eggs Retrieved	# Eggs Fertilized	Did a pregnancy occur?	Did a live birth occur?

Were their embryos left to cryopreserve (freeze)? N/A If yes, approximately how many per cycle? N/A

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

What is the least amount you would consider? \$5,000

Currently I have a career in: Full-Time Student

I have been in this profession for 3 days/mos/years

Will you require missed wages from work? No

If yes, what is your hourly wage? N/A

How many hours per week do you work? N/A

Are you eligible to work in the United States? Yes

Is your work schedule flexible? N/A

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? ☒ Yes \_\_\_\_ No

## MEDICAL/SOCIAL HISTORY

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

If yes, please explain: N/A

Have you ever had any surgeries? If so please list type and date:

1. 08/21/2008-Right knee surgery- tore my ACL from lacrosse game
2. 06/18/2012-Cornea Eye Transplant from being hit in the eye with a lacrosse ball

Have you had any serious illness in the past? No

If yes, please describe: N/A

Have you had a blood transfusion in the last 12 months? No

If yes, please list date and reason: N/A

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

Any hospitalizations not mentioned above? No If yes, please explain: N/A

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

Please explain: None

Have you been exposed to excess radiation or toxic chemicals in your work or personal life? No

Have you ever had a reaction to anesthesia? No If yes, please explain reaction in detail: N/A

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

\*Do you smoke cigarettes? No      Packs per day?                 # of years                 # of years quit           

Do you now or have you ever taken recreational drugs? No If so, What? \_\_\_\_\_

Do you drink alcohol? No If yes, how many drinks per: day? \_\_\_\_\_ week? \_\_\_\_\_ month? \_\_\_\_\_

Do you have any allergies to drugs or environmental exposures? No Pls. explain: \_\_\_\_\_

Describe any childhood allergies that you have outgrown: Pollen

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

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

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

**\*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.**

Do you have any medical illnesses (diabetes, asthma, etc...)? No If yes, pls. explain: \_\_\_\_\_

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

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?
11/22/2009	Half human heart and strawberry	Back neck	Yes
11/22/2011	Fly	Wrist	Yes

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

<i>Medication</i>	<i>How Often</i>	<i>Reason</i>
<u>Pred Forte</u>	<u>2x a day</u>	<u>Eye drops</u>
_____	_____	_____

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

<i>Medication</i>	<i>How Often</i>	<i>Reason</i>
_____	_____	_____
_____	_____	_____

**Religion Born Into:** Catholic

**Religion Practiced:** Catholic/Buddhism



## EDUCATION

H.S. Grade Point Average (GPA): 3.5      SAT Scores: Verbal \_\_\_\_\_ Math \_\_\_\_\_      ACT Score: 32

Education:      \_\_\_\_\_ Did not Complete High School  
                     \_\_\_\_\_ Received GED  
                     ☒ Completed high school  
                     \_\_\_\_\_ Completed/enrolled in Vocational Schooling \_\_\_\_\_  
                     ☒ Currently in college, pursuing degree in Nursing/Psychology  
                     \_\_\_\_\_ Completed college, degree in \_\_\_\_\_ GPA: \_\_\_\_\_  
                     \_\_\_\_\_ 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 Colorado in Colorado Springs</u>	<u>2011-2015</u>
2.	_____	_____

What was your favorite subject in school? History      You're least favorite? Math

Dean's List or Honor Roll? Both

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

Academic Strengths (i.e. math, reading): Reading and English

Other skills or talents? \_\_\_\_\_

Do you show artistic or musical ability? Yes If yes please explain: Piano, Violin and Saxophone

Languages:    Speak: English  
                     Read: English  
                     Write: English

## HEALTH HABITS

Exercise Habits: \_\_\_\_\_None \_\_\_\_\_Occasional ☒ Regular

Physical activities include: Lacrosse, soccer and martial arts

Have you excelled in any physical activities? ☒ Yes \_\_\_\_\_ No

If Yes, What: Lacrosse, soccer, martial arts

Manual Dexterity:    Dexterous    (Average)    Clumsy

Your diet is: \_\_\_\_\_Vegetarian ☒ Non-vegetarian \_\_\_\_\_Vegan      Your diet is:    poor    average    (excellent)

Do you have any dietary restrictions? None

## FAMILY HEALTH HISTORY

	Natural Hair Color	Eye Color	Height Weight	Skin Tone	Age If Living	Age at Death	Cause of Death
Mother	Black	Brown	5'02",118	tan	47		
Father	Brown	Green	5'10",210	light	52		
Brother: 1.	Black	Hazel	5'10", 160	tan	19		
2.							
3.							
4.							
Sister: 1.							
2.							
3.							
4.							
Maternal Grandmother	Black	Brown	5'01",110	Tan	72		
Maternal Grandfather	Brown	Brown	5'05",160	Tan	80		
Paternal Grandmother	Red	Green	5'05",110	white	72		
Paternal Grandfather	Blonde	Green	5'10" 210	white		70	Natural
Children: (If Any) 1.							
2.							
3.							
4.							

Are you adopted? No If yes, do you have access to your biological health history? N/A

Twins or multiple births in the family? No If yes, how many sets? N/A

## GENETIC HISTORY

Are there any known genetic diseases that run in your family? No If yes, please identify all such diseases and explain in as much detail as possible: N/A

Has anyone in your family been born with a birth defect? No If yes, please explain in detail: N/A

Have you had a brother or sister die in infancy or early childhood? No If yes, please explain the cause of death: N/A

Are there any members of your family with a history of learning disabilities or autism? No  
If yes, please explain: N/A

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

Have you had a chromosomal analysis performed? No If so, were your results of normal XX karyotype? N/A

Is there anything else we should know about your family?  
Not at this time

## PERSONAL AND MOTIVATIONAL

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

I would have to say in my own words my personality is very optimistic, outgoing, very out spoken, independent, and head strong. I rarely ever have a temper; it takes a lot to make me get angry, I believe there is no need to get all worked up over anything; things happen that's part of life. We learn from the mistakes we make so no need to be mad and have a temper. My character is very bubbly, smart, good head on my shoulders, good morals, high self esteem, always looks at the bright things in life. There's always a reason to smile.

### **What is your philosophy of life?**

Life is a beautiful thing so cherish what you have and never take for granted the people who we are blessed to come across. Always look at the future with an optimistic attitude and always learn from the mistakes we make.

### **What qualities and characteristics would you hope the recipient parents possess?**

I would hope the recipients possess good hearts and good morals on life. Two parents who are very easy going but at the same teaches things that are meant to be taken serious in life. And to always be there to fall back on if something was wrong. To always be there for the child, be a best friend but be a parent as well.

### **How does it make you feel at the possibility of their offspring knowing about the donation?**

It makes me feel like I did something amazing for two parents who weren't able to conceive and I'm totally okay with the offspring knowing because they should know they were brought in this world for a reason. They made a difference in someone's life.

### **How would you describe your childhood?**

I had an amazing childhood, my parents made sure we had everything we needed from a good education, to all the toys in the world but they also made sure we knew what love was and what was right from wrong. My parents always let us try something new as a child, we were able to learn how to play instruments but we were also able to learn every sport we wanted to try. They always made sure we ate dinner as a family and always took time out there day to ask us about ours.

### **What is the earliest memory you hold as a child?**

The earliest memory I hold as a child would have to be Christmas when I was 8 years old. I will never forget that Christmas because that was the Christmas my dad finally decided to buy me a kitten. All I ever wanted was a cat and my dad would always say no because he was allergic so when I finally gave up on having a cat, that Christmas I came downstairs and there was my dad with a kitten in his arms with a pink bow and a collar I picked out for one of my toys and he gave her to me and said this is your kitten, take care of her, and love her. I then asked about my dad's allergy and he said he went to the doctors and got medicine to help him cope with being around a cat. Till this day I still have that very cat and she has done nothing but make me happy and that's all thanks to my dad.

### **What was it like growing up in your family?**

My family is pretty much amazing. I couldn't have asked for better parents and a better baby brother. My parents made sure they taught us everything we needed to know about life. They always made sure to give us a hug and a goodnight kiss. They always talked to us about our days and our problems. They were our support. My parents always told us to never give up on something we believe in even if we fall down all that means is get back up and try harder. As I grew up I became best friends with my parents and my brother. They are the only people I trust and can always count on to be there for me. They never give up on me even when I have. Growing up in my family is a blessing and I am very thankful to have a family like mine.

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

If I could change one thing about me it would have to be trying to be not so much a perfectionist. To just let things happen the way they're supposed to be and not so much try to make them perfect. I would change that because not everything in this world can be perfect. Flaws make things beautiful and something we all need to accept, life isn't perfect.

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

I admire my mom. The reason why I admire my mom out of anyone in this world is because she is the person who taught me to a strong independent woman. She is the person who wiped my tears from my first heartache to the same person who helped me get all dolled up for my first date, My mom is the person who taught me to love myself and to never let anyone tell me differently or to put me down. She has always been there for me through everything from teaching to bake my first cake to teaching me about makeup and hair. I am not the person I am today if it wasn't for her.

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

If I could do anything I wanted on a perfect day it would have to be swimming with sharks and then riding every ride at Disney World!

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

As a child I had a very out-spoken personality; I always had to know and be friends with every child in my class. I was always smiling and outgoing. I was a ball of joy. I don't believe I ever had a temper as a child. I was always taught to communicate your feelings through talking it out, never hit or scream your feelings.

**When I Was A Child:**

My favorite thing to do was: play house

At home I was expected to: have manners

My parents were strict about: grades

My parents taught me to value: the little things

What I loved most about my father was: that he was always our biggest supporter

What I loved most about my mother was: that she always gave us a hug and said I love you after everything

My favorite relatives were: grandma and grandpa

I loved to visit: the zoo especially; the lions and tigers

In comparison to others I was: Bubbly

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

	Intellectual/Academic Achievements	Artistic Achievements
Mother	Honor roll	Piano and Violin
Father	Honor roll/ Deans list	
Sisters		
Brothers	Honor roll/presidential award	Draws and paints.

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

My personality as a teenager didn't really differ from when I was a child. I always made sure I knew everyone in my class or on my team. I was very outgoing and always spoke what was on my mind, kind and sweet, always lent a hand when it was needed. I also can say I have a great sense of humor. I never had a temper in high school, I had no need for one.

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

None

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

The most important influence on me would had to have been my lacrosse coach. She influenced me to go so much further in my skills in lacrosse. She always pushed me to do better and always told me what was right from wrong. She is the main reason why I took my grades in school so serious because if you failed you couldn't play on the team. She always influenced me to push myself for greater and to never settle for anything less.

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

My ambitions and goals as a teenager were to go to college and to major in nursing, to make sure I knew everyone in my graduating class and of course to be prom queen.

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

My best subjects in school were English, history and writing. My worst subjects in school were math and Spanish.

**What do you hope to achieve by volunteering in an egg donor program?**

To make a difference in someone's life.

**What helped you decide to become an egg donor?**

No one just something I heard about and decided on my own to do.

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

I wish the best for you and your family and I really hope your dreams come true.

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

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

**Would you be willing to donate to gay or single prospective parents? Yes**

**Please specify:** It doesn't matter who the parent is as long as there good people.

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

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

**Would you describe yourself as a religious or spiritual person?** A little bit of both.

**Do you have any ethical, moral or religious reservations about being an egg donor?** None

**What are your personal goals? Have you achieved any of these goals?**

My personal goals are to make a difference in at least one person's life and to at least be able to travel the world. If I become an egg donor then yes I would be achieving my goals.

**What do you see yourself doing in the next 5-10 years?**

I see myself living in California and attending medical school and at the same time working as an RN

**What would you like your recipient couple to know about you that has not already been asked?**

Nothing unless they have something else they want to know more about.

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

I am okay with aborting a fetus depending on what the defect is.

**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?**

Yes

**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?**

No

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

I'm ok with that.

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

I'm ok with that.

**What is your favorite color?** Pink

**Favorite type of food?** Italian

**Favorite movie?** Beauty & the Beast

**Favorite type of music?** Alternative

**Favorite Book?** Harry Potter series

**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?**

Hi there,

I want you to know you were brought to this world to help a couple that was un able to conceive on their own. You are what we would call a miracle and you are making a set of parents very happy and just know you made a difference in someone's life. It is a blessing to know just from one small act came a huge outcome. The parents you have so badly wanted you and cherish just having the ability to have you as their own.

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**



<b>HEART</b>	<b>You</b>	<b>Mother</b>	<b>Father</b>	<b>Siblings</b>	<b>Grandparents</b>	<b>Other Family</b>	<b>Explanation (which side of family, age of onset, etc.)</b>
A. Stroke	No	no	no	no	no	no	
B. heart attack	No	no	no	no	no	no	
C. heart disease	No	no	no	no	no	no	
1. from birth	No	No	No	No	No	No	
2. lifestyle	No	No	No	No	No	No	
D. hardening of the arteries	No	No	No	No	No	No	
E. high blood pressure	no	No	No	No	No	No	
<b>BLOOD</b>							
A. anemia	No	No	No	No	No	No	
B. sickle-cell anemia	No	No	No	No	No	No	
C. hemophilia or other bleeding problem	No	No	No	No	No	No	
D. leukemia	No	No	No	No	No	YES	Uncle from my dad's side, he was 28 and passed away at 32
E. Immune Deficiency	No	No	No	No	No	No	
F. Factor V Leiden thrombophilia (Blood clots or strokes)	No	No	No	No	No	No	
G. other blood disorder	No	No	No	No	No	No	
<b>RESPIRATORY (LUNGS)</b>							
A. hay fever	No	No	No	No	No	No	
B. asthma	No	No	No	No	No	No	
C. emphysema	No	No	No	No	No	No	
D. tuberculosis	No	No	No	No	No	No	
E. lung cancer	No	No	No	No	No	No	
F. pneumonia	No	No	No	No	No	No	
G. Alpha-1 antitrypsin Disorder	No	No	No	No	No	No	
H. other lung disease	No	No	No	No	No	No	
<b>GASTRO-INTESTINAL</b>							
A. ulcer of stomach or duodenum	No	No	No	No	No	No	
B. gall stones	No	No	No	No	No	No	
C. hepatitis A,B or C	No	No	No	No	No	No	
D. appendicitis	No	No	No	No	No	No	
E. cirrhosis	No	No	No	No	No	No	
F. colon cancer	No	No	No	No	No	No	
G. ulcerative colitis	No	No	No	No	No	No	
H. Crohn's disease	No	No	No	No	No	No	
I. cystic fibrosis	No	No	no	No	No	no	
K. pyloric Stenosis	No	No	No	No	No	No	
L. multiple Polyps of the Colon	No	No	No	No	No	No	
m. rectal Disorder	No	No	No	No	No	No	
n. inflammatory Bowel Disease	No	No	No	No	No	No	
o. any other problem of the digestive system	No	No	No	No	No	No	
<b>METABOLIC/ENDOCRINE</b>							
A. diabetes mellitus	No	No	No	No	No	No	
B. hypoglycemia	No	No	No	No	No	No	
C. thyroid cancer	No	No	No	No	No	No	
D. thyroid disease	No	No	No	No	No	No	
E. goiter	No	No	No	No	No	No	
F. adrenal dysfunction or disorder	No	No	No	No	No	No	
G. hyperactivity	No	No	No	No	No	No	
H. lumps or cysts in breast	No	No	No	No	No	No	
I. polycystic ovarian syndrome	No	No	No	No	No	No	
J. pelvic inflammatory disease (PID)	No	No	No	No	No	No	
K. endometriosis	No	No	No	No	No	No	

	You	Mother	Father	Siblings	Grandparents	Other Family	Explanation (which side of family, age of onset, etc.)
<b>URINARY</b>							
A. kidney disease	No	No	No	No	No	No	
B. other disease of urinary tract (urethra, bladder, ureter)	No	No	No	No	No	No	
<b>GENITAL/REPRODUCTIVE</b>							
A. undescended testicle	No	No	No	No	No	No	
B. hypospadias	No	No	No	No	No	No	
C. prostate cancer	No	No	No	No	No	No	
D. uterine fibroids	No	No	No	No	No	No	
E. ovarian cysts	No	No	No	No	No	No	
F. cancer of the cervix, ovaries or uterus	No	No	No	No	No	No	
<b>NEUROLOGICAL</b>							
A. migraines	YES	No	No	No	No	No	I get migraines maybe once every two months started getting them when I was 18
B. mental retardation	No	No	No	No	No	No	
C. senility before age 50	No	No	No	No	No	No	
D. Multiple Sclerosis	No	No	No	No	No	No	
E. Cerebral Palsy	No	No	No	No	No	No	
F. Neurofibromatosis	No	No	No	No	No	No	
G. ADHD	No	No	No	No	No	No	
H. Autism/Asperger's	No	No	No	No	No	No	
I. Tuberous sclerosis	No	No	No	No	No	No	
J. Parkinson's disease	No	No	No	No	No	No	
K. Scoliosis	No	No	No	No	No	No	
L. Myasthenia Gravis	No	No	No	No	No	No	
M. Tourette's Syndrome	No	No	No	No	No	No	
N. epilepsy/seizures	No	No	No	No	No	No	
O. hydrocephalus	No	No	No	No	No	No	
P. disorder of the spinal cord	No	No	No	No	No	No	
Q. Huntington's chorea	No	No	No	No	No	No	
U. Gaucher's disease	No	No	No	No	No	No	
R. Wilson's disease	No	No	No	no	No	No	
S. Creutzfeldt-Jacob disease	No	No	No	no	No	No	
T. Alzheimer's disease	No	No	No	No	No	No	
U. other diseases of the nervous system	No	No	No	No	No	No	
<b>MENTAL HEALTH</b>							
A. schizophrenia	No	No	No	No	No	No	
B. bipolar or manic depressive	No	No	No	No	No	No	
C. depression	No	No	No	No	No	No	
D. anxiety/panic attacks	No	No	No	No	No	no	
E. anorexia/bulimia/other eating disorder	No	No	No	No	No	No	
F. suicide attempts	No	No	No	No	No	No	
G. other mental health disorder requiring hospitalization	No	No	No	No	No	No	
<b>MUSCLE/BONE/JOINTS</b>							
A. muscular dystrophy	No	No	No	No	No	No	
	No	No	No	No	No	No	
C. lupus	No	No	No	No	No	No	
D. deformity of the spine	No	No	N	O	No	No	
E. osteoporosis	No	No	No	No	No	No	
F. dwarfism	No	No	No	No	No	No	
	<b>You</b>	<b>Mother</b>	<b>Father</b>	<b>Siblings</b>	<b>Grandparents</b>	<b>Other Family</b>	<b>Explanation (which side of family, age of onset, etc.)</b>

<b>Cont.</b>							
G. heredity low back disease	No	No	No	No	No	No	
H. arthritis	No	No	No	No	YES	No	My grandma on my dad's side has arthritis, she got it when she was 68
I. gout	No	No	No	No	No	No	
J. Osteogenesis imperfecta (brittle bone disease)	No	No	No	No	No	No	
K. loss of muscle coordination	No	No	No	No	No	No	
L. Marfan syndrome	No	No	No	No	No	No	
M. spinal muscular atrophy	No	No	No	No	No	No	
N. Reiter's disease	No	No	No	No	No	No	
O. myasthenia gravis	No	No	No	No	No	No	
P. metabolic bone disease (be more specific)	No	No	No	No	No	No	
<b>SIGHT/SOUND/SMELL</b>							
A. deafness before age 60	No	No	No	No	No	No	
B. deformity of the ear	No	No	No	No	No	No	
C. cataracts before age 50	No	No	No	No	No	No	
D. blindness	No	No	No	No	No	No	
E. color blindness	No	No	No	No	No	No	
F. glaucoma	No	No	No	No	No	No	
G. deviated septum	No	No	No	No	No	No	
H. retinoblastoma	No	No	No	No	No	No	
I. retinitis Pigmentosa	No	No	No	No	No	No	
J. any other sight/sound/smell disorders	No	No	No	No	No	No	
<b>SKIN</b>							
A. acne	No	No	No	No	No	No	
B. eczema	No	No	No	No	No	No	
C. skin cancer	No	No	No	No	No	No	
D. pigmentation disorders	No	No	No	No	No	No	
E. excessive facial hair	No	No	No	No	No	No	
F. psoriasis	No	No	No	No	No	No	
G. neurofibromatosis	No	No	No	No	No	No	
H. infectious skin disease	No	No	No	No	No	No	
I. other disorders of the skin	No	No	No	No	No	No	
<b>CONGENITAL ABNORMALITIES/BIRTH DEFECTS</b>							
A. cleft lip or palate	No	No	No	No	No	No	
B. congenital hip problems	No	No	No	No	No	No	
C. club feet	No	No	No	No	No	No	
D. heart defect	No	No	No	No	No	No	
E. hearing problems	No	No	No	No	No	No	
F. Spina bifida-neural tube (open spine)	No	No	No	No	No	No	
G. Microcephaly	No	No	No	No	No	No	
H. holoprosencephaly-a single-lobed brain structure and severe skull and facial defects	No	No	No	No	No	No	
I. other	No	No	No	No	No	No	
<b>CHROMOSOMAL ABNORMALITIES</b>							
A. down syndrome B. other (i.e. Turner, Fragile X, Klinefelter's etc..)	No	No	No	No	No	No	

OTHER							
Cont.	You	Mother	Father	Siblings	Grandparents	Other Family	Explanation (which side of family, age of onset, etc.)
A. alcoholism	No	No	No	No	No	No	
B. drug abuse, misuse or addiction	No	No	No	No	No	No	
C. breast cancer	No	No	No	No	No	No	
D. any other cancer not mentioned above	No	No	No	No	No	No	
E. any other condition not mentioned above	No	No	No	No	No	No	

## RISK FACTORS

Yes

No

Comment

Have you ever been sexually active with a male who was gay or bisexual?

Yes

(No)

Have you ever injected drugs or had a sexual partner who did so?

Yes

(No)

Have you ever had hemophilia or received any human derived clotting factor concentrates, including factor VIII or factor IX concentrate?

Yes

(No)

Have you ever had a sexual partner with hemophilia or who received any human derived clotting factor concentrates?

Yes

(No)

Have you ever had sex in exchange for money or drugs?

Yes

(No)

Have you ever been sexually active with a person who has had sex in exchange for money or drugs?

Yes

(No)

Have you ever been sexually active with a person who was known or suspected to have HIV, hepatitis B or hepatitis C?

Yes

(No)

Have you been exposed to body fluids, open wounds, non-intact skin or mucus membranes of any person known or suspected to have HIV, hepatitis B and/or C?

Yes

(No)

**(Cont'd)**

Have you had an accidental needle stick within the past 12 months?      **Yes**      **(No)**

---

Have you ever been or have you had a sexual partner who was incarcerated for 72 consecutive hours or longer?      **Yes**      **(No)**

---

In the past 12 months, have you lived with or had contact with anyone known or suspected to have hepatitis?      **Yes**      **(No)**

---

Have you acquired a tattoo or other skin piercing procedure within the preceding 12 months?      **Yes**      **(No)**

---

Have you ever been diagnosed with hepatitis?      **Yes**      **(No)**

---

Have you been vaccinated or had contact with anyone vaccinated for smallpox within the past 2 months?      **Yes**      **(No)**

---

Have you ever been diagnosed with or suspected to have West Nile Virus?      **Yes**      **(No)**      **if so, when?**

---

Have you ever been treated for or diagnosed with chlamydia, gonorrhea, herpes or syphilis?      **Yes**      **(No)**      **if so, when?**

---

Have you or any of your blood relatives been diagnosed and/or have a history of transmissible spongiform encephalopathy such as Creutzfeldt-Jakob disease or variant Creutzfeldt-Jakob disease?      **Yes**      **(No)**      **if so, who?**

---

Have you ever received a non-synthetic dura mater transplant or a pituitary-derived growth hormone?      **Yes**      **(No)**

---

Do you have a history of changes in cognition, speech or gait?      **Yes**      **(No)**

---

Have you ever received a blood transfusion?      **Yes**      **(No)**      **if so, where?**

---

**(Cont'd)**

Have you visited or lived in the United Kingdom for three months or more between 1980-1996 including England, Scotland, Wales, Ireland, Isle of Man, Channel Islands, Gibraltar or Falkland Islands?      **Yes**      **(No)**

---

Were you a member of the US military, civilian military, employee or a dependent of a member of the military stationed in Belgium, the Netherlands, Germany, Spain, Portugal, Turkey, Italy or Greece between 1980-1996?      **Yes**      **(No)**

---

From 1980 to present, have you spent time that adds up to 5 years or more in Europe?      **Yes**      **(No)**      **if so, where?**

---

Were you born in or have you lived in any of the following Countries since 1977; Cameroon, Central Africa Republic, Chad, Congo, Equatorial, Guinea, Gabon, Niger or Nigeria?      **Yes**      **(No)**      **If so, when?**

---

If yes, were you given a blood transfusion or any medical treatment with a product made from blood while you were there?      **Yes**      **(No)**

---

Have you ever had sexual contact with anyone who was born or lived in Cameroon, Central Africa Republic, Chad, Congo, Equatorial, Guinea, Gabon, Niger or Nigeria since 1977?      **Yes**      **(No)**

---

Have you or someone you know been diagnosed, treated or suspected of having sudden acute respiratory syndrome? (SARS)?      **Yes**      **(No)**      **if so, when?**

---

Have you, your sexual partner, and/or anyone you live with ever had a transplant or other medical procedure that involves being exposed to live cells, tissues or organs from an animal?      **Yes**      **(No)**      **if so, who?**

---

Have you been exposed to blood, saliva or fluids from the person described in the proceeding question?      **Yes**      **(No)**

---

(Cont'd)

Have you ever received a human organ, tissue ☒ **Yes** ☐ **No**  
transplant or human extract? *I received a cornea eye transplant from getting hit hard in the eye by a lacrosse ball which caused nerve damage so my eye couldn't heal on its own. 06/18/2012*

---

Have you ever been excluded as a blood donor? **Yes** ☒ **No** if so, why?

---

Have you been diagnosed or suspected to have Chagas' disease? **Yes** ☒ **No**

---

Have you been exposed to significant levels of radiation, toxic chemicals, or heavy metals (such as lead, mercury or gold) in your home or work environment? **Yes** ☒ **No**

---

Have you received a bite from an animal suspected for rabies within the last six months? **Yes** ☒ **No**

---

### CONSENT

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: 02/02/2013

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: 02/02/2013

Witness to Signatures above: \_\_\_\_\_

Date: 02/02/2013

## 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.
- **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.
- **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.
- **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

## 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.

- **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.

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