The Case for Comprehensive Medical and Genetic Testing of Gamete Donors

Donor Sibling Registry

About the DSR & its Membership

The Donor Sibling Registry (DSR) is a non-profit, worldwide organization dedicated to educating, connecting and supporting donors, recipients and offspring. With more than 29,800 members, the DSR has helped to connect more than 8,100 half siblings and/or donors with each other.

The DSR doesn't just generate genetically related joy- it also shines light on serious genetic concerns about gamete donation. Frequently, the DSR counsels recipients whose children have inherited undisclosed genetic disorders, or who have discovered that their donor was dishonest regarding health, or that the sperm bank didn't notify them about a reported illness.

US donors can father many offspring (at least one US donor is known to have fathered more than 125 offspring, so far) so a greater number of people will be at risk from a single person's genetic makeup.

Sources of Information

Medical and genetic information compiled from surveys, direct reporting to the DSR, and as reported by Cabri DNA testing.

2008 Survey - 155 Egg Donors

2009-Survey - 164 Sperm Donors

2009 Survey - 759 Donor Offspring

2009 Survey – 1700 Sperm Donor Recipients

Medical Update Requests from Clinics and Sperm Banks, Donors with Medical and Genetic Issues to Share, Sperm Donors Would Accept Genetic Testing

Currently, many US facilities either refuse to update donor/offspring medical information, or even if they accept updates, refuse to share the information, or make the process of reporting so complex and expensive that donors and recipients simply cannot comply or afford it. In addition, US sperm banks do not have an accurate accounting of all children born from any one donor, so if illness is reported, it is then impossible to notify all relevant families.

84% of sperm donors have **never been contacted** by their clinic(s) for medical updates.

96% of egg donors have **never been contacted** by their clinic(s) for medical updates.

23% of sperm donors felt that they **had medical/genetic issues** that would be important to share with families.

31% of egg donors felt they **had medical/genetic issues** that would be important to share with families.

94% of sperm donors would have **accepted an offer for genetic testing**, had it had been offered by their sperm banks.

Privately Reported Health & Genetic Issues from Donors

Medical and genetic issues reported by sperm and egg donors for themselves or their immediate family include:

Albinism, Alcoholism, Aspergers, Autism, Bi-Polar Disorder, Brain Aneurysm, Breast Cancer, CF Carrier, Canavan Disease, Cavernous Angioma, Colon Cancer, Congenital Heart Disease, Hashimoto's Syndrome, Hemachromatosis, High Blood Pressure Leading to Stroke, Leukemia, Lung Cancer, Melanoma, Mitral Valve Prolapse, Multiple Myeloma, Multiple Sclerosis, Polycystic Kidney Disease, Prostate Cancer, Rheumatoid Arthritis, Spinal Muscular Atrophy, Type II Diabetes, Ulcerative Colitis

Privately Reported Health & Genetic Issues from Recipients and Offspring

Acute Lymphoblastic Leukemia, ADD, ADHD, Albinism, Amniotic Band Syndrome, Aspergers, Asthma, Atrial Septal Defect, Auto Immune Thyroiditis, Bi-Polar Disease, Branched-chain Ketoaciduria, Complex Congenital Heart Defect, Congenital Heart Disease, Congenital Hypothyroidism, Cystic Fibrosis, Dwane Syndrome, Ebsteins Anolomy, Ectodermal Dysplasia, Heart Murmur, Hemoglobin D, Hemophagocytic Lymphohistiocytosis, Hole in Heart, Horseshoe Kidney, Hydrocephalus, Hypertrophic Cardiomyopathy, Hypophosphatasia, Hypospadias, Imperforated Anus, Juvenile Dermatomyositis, Juvenile Arthritis, Karatosis Pilaris, Kidney Disease, Lethal Dwarfing Syndrome, Marfan's Syndrome, Medium Chain Acyl CoA Dehydrogenase Deficiency (MCAD), Metabolic Genetic Disorder, Mitral Valve Stenosis, Multiple Hereditary Exostosses, PANDAS, PHACES Syndrome, Phenylketonuria (PKU), Polycystic Kidney Disease, Prader-Willi, Rasmussen's Encephalitis,Renal Disease, Retinoblastoma, Seizure Disorders,Severe Congenital Neutropenia, Spinal Muscular Atrophy, Tay Sachs, Tourettes, Tracheo-Esophogeal Fistula, Truncus Arteriosis, Type I Diabetes, Van Der Woude Syndrome, Vesicoureteral Reflus, Von Willebrand Disease, Williams -Beuren Syndrome, Zellweger Syndrome

See Attachment for additional newly reported genetic and medical issues.

Offspring Who Desire Contact with Donors for Genetic/Medical Information

74% of donor offspring who wish to make contact with their donors list learning more about their medical background as a main reason for the desired contact.

Publicly Reported Health & Genetic Issues

2011: At least nine children (Nordic Cryobank & California Cryobank) that have been conceived with the sperm of a man with a genetically inherited disorder Neurofibromatosis (NF1 or von Recklinghausen disease). *Thelocal.com.*.
2009: London Women's Clinic used chromosomally abnormal donor sperm to treat 11 women including a couple who had to destroy 22 embryos created over a year of treatment. *The Independent* and *BioNews*.
2009: A child conceived using gametes from anonymous sperm and ova donors was diagnosed with spinal muscular atrophy type 1. *Fertility and Sterility*
2009: New England Cryogenic sued by a woman claiming that her children inherited genetic disorders. Other families who used this donor also report issues. *Boston Herald*.
2009: A Pacific Reproductive Services donor passed along HCM, a fatal heart disease to 9 of his 22 known offspring. One child consequently died. *J Am Med Assn*.
2008: Fragile X inherited from an Idant Lab. donor. *The Am. J Med Genetics*.
2008: Two donor sibling cohorts from California Cryobank have a very high percentage of their children diagnosed with PDD-NOS (Autism). *O Mag*.
2006: An International Cryogenics donor transmits Severe Congenital Neutropenia to at least 5 offspring. J Pediatrics.
2006: A Fairfax donor offspring diagnosed with Delta Storage Pool Deficiency (delta-SPD). *SELF Mag.*
2004: A Fairfax donor transmitted familial Hemophagocytic Lymphohistiocytosis (FH), to twins, one child subsequently died. *SELF Mag.*
2002: A Dutch donor conceived 18 children before being diagnosed with Autosomal Dominant Cerebellar Ataxia (ADCA). *J. Med. Ethics*.
2002: One recipient, two cases of spinal muscular atrophy (SMA). *Fertility and Sterility*
2001: British based donor of Australian origin with at least 43 offspring, passed along potentially fatal genetic disorder -Opitz Syndrome. *London Sunday Times*.
1995: California Cryobank donor who donated 1500 vials of sperm transmitted Autosomal

Current Practices & Regulation

In the US, FDA oversight has been directed at the prevention of infectious diseases including STD's. Little attention has been paid to the potential transmission of genetic diseases. (US & UK requirements are similar.)

Current US Screening:

Sexually Transmitted Diseases: HIV, HTLV, Hepatitis B & C, Syphilis, Gonorrhea, Chlamydia, CMV

Genetic testing varies significantly at US clinics as adhering to ASRM recommendations is voluntary.

The less screening carried out, the fewer donors need be disqualified. Fewer tests also equals less cost.

Some clinics and sperm banks test for some of the following:

Current US Genetic Testing (Select Groups only):

Cystic Fibrosis, Sickle-Cell Disease, Tay Sachs, Canavan Disease, Gauchers Disease, Nieman-Pick's Disease, B-Thalassemia

Recommended Medical & Genetic Testing

Testing for ALL donors: Karyotyping, Cystic Fibrosis, Tay Sachs, Fragile X, Hemachromatosis (for HFE mutation), BRCA 1 & 2, Celiac Disease, Polyposis Conditions Caused by Mutations in the APC Gene, Hereditary Non-Polyposis Colorectal Cancer (HNPCC), Glycogen Storage Diseases such as Fabry's and Niemann-Pick, Polycystic Disease, Huntington's Disease, Melanoma (CDKN2A Gene) and Myopia-Eyesight. Marfan's testing for donors over 6'2".

Additionally: More thorough physical examinations including organ function, a face-to-face medical history intake and full psychological screening.

Other Recommendations

*Periodic donor quarantine using the "5&2" protocol. A donor would be permitted no more than 5 pregnancies. They would then be quarantined until the youngest reached age two and completed an extensive pediatric evaluation, along with the other 4 half siblings.

*Track all recipients, donors and births.
*Mandatory reporting of all live births from each donor.
*Limit the number of births conceived form any one donor.

*Require donors to regularly update their family medical history and have this information available to all families who have used this donor.

*Encourage donors, parents and offspring to share and update medical and genetic information with each other.

*Require legal and financial protection for all donors so that they may feel safe to update and report medical issues.

*Consequences for donors who knowingly withhold important medical and genetic information from clinics

*Counsel parents on openness, full disclosure and the importance of using open donors. Emphasize the importance of people having information about their genetic, ancestral and medical backgrounds. Counsel all donors on the same.

*Initiate follow-up health history reporting of egg donors.

2010
Additional Genetic and Medical Issues (not known to be present in recipient's family) reported in Donor Sibling Registry Surveys for Parents and privately reported:

Agenesis of the Corpus Callosum
Alpha 1 Deficiency
Alpha Thalassemia Trait
Apraxia
Arnold-Chiari malformation
Atrial Septal Defect (ACD)
Bi-cuspid Aortic Valve Disease
Borderline Personality Disorder
Cardiac (ASD PDA) and Pulmonary Hypertension
Cerebral Palsy
Chromosome abnormality:5p minus or Cri-du-chat.
Coarctation of Aorta
Congenital Lobar Emphysema
Cornelia de Lange Syndrome
Craniosynostosis
Cystic Hygroma
Dandy Walker Variant
Depression
Down Symdrome
Dysgraphia
Eosinophilic Esophagitis
Epilepsy
Febrile Seizures
Femoral Antiversion
Fragile X
Genetic High Cholesterol
Goldenhar Syndrome
Graves Disease
Hashimoto's disease (chronic lymphocytic thyroiditis)
Hydrocephalus
Hypotonia
Ileal Atresia
Kidney Reflux Stage 3
Kleinfelter's Syndrome
Langerhans Cell Histiocytosis
Leukemia
Marcus Gunn Syndrome
Medulloblastoma Brain Tumor
MTHFR C677T gene mutation
Myelomeningocele (Spina bifida)
Myotonic Muscular Dystrophy (MMD)
Neublastoma Stage 3
Neurofibromatosis Type 1
NUT Midline Carcinoma
Oppositional Defiant Disorder
PCOS
PDD-NOS
Pectus Excavatum
Plagiocephaly
Pyloric Stynosis
Primary Sclerosing Cholangitis
Radioulnar Synostosis
Rett Syndrome
Robersonian Translocation Chromosome
Rotary Nystagmus
Reitters Syndrome
SVT- Superventricular Tachycardia
Sagittal Craniosynostosis
Scoliosis
Sensory Integration Dysfunction
Sickle Cell Carrier
Sieves Disease
Spastic Quad Cerebral Palsy
Strabismus
Sub-Aortic Membrane
Tethered Spinal Cord Syndrome
Third Degree Heart Block
Thyroid Cancer
Torticollis
Trachea Esophogeal Fistuala
Vacteral Association
Vasovagal Syncope
Vesico-Uretal Reflux
Wilm's Tumor (Kidney Cancer)
Wolff-Parkinson-White Syndrome
Zellweger Synndrome