

FRIENDS OF THE MICHAEL FUND NEWSLETTER

Fall – Winter 2003

Web Page www.michaelfund.org

The Michael Fund/International Foundation for Genetic Research

4371 Northern Pike, Pittsburgh, PA 15146 Phone 412-374-0111

Happy 25th Anniversary to the Michael Fund

This is our special 25th anniversary issue of the Michael Fund Newsletter. It is our way of expressing our appreciation to the many individual donors, organizations and foundations that have made our pro-life genetic research program possible over the last two and a half decades. Thank you all and God bless.

Randy Engel, Editor

Like all organizations, the International Foundation for Genetic Research popularly known as the Michael Fund began with an idea and an ideal.

The idea to create a pro-life genetic research agency came from Dr. Hymie Gordon of the Mayo Clinic in 1977. The ideal was based on the therapeutic genetics research program of Dr. Jerome Lejeune, Professor of Fundamental Genetics at the University of Paris and the discoverer of the extra-chromosome that causes Down syndrome (Trisomy 21).

In 1978, Randy Engel, Director of the U.S. Coalition for Life contacted Dr. Lejeune at his Paris home and shared Dr. Gordon's idea with him. Dr. Lejeune thought the phone call almost miraculous since his grant from the French government was coming to an end and he did not know where the money was going to come from to continue his research to find a cure or treatment for Down syndrome and related chromosomal disorders.

Dr. Lejeune has always believed that there is no reason why chromosomal disorders that result in mental deficiencies cannot be cured.

"There is no reason to believe that nothing can be done for mentally retarded children," he said. "But research into the causes and cures for mental deficiency must have *IMPETUS*...If we continue to 'treat' mentally defective children by aborting them, we will never be in a position to cure them, to give them back their intelligence," he said.

"Instead of the National Institute of Death that abortionists try to make medicine into, we have to build an International Institute of Life which will try to tell scientists of the world that they have an enormous task which is to try to alleviate the difficulties of these children..." Dr. Lejeune concluded.

Naturally, that very night, Dr. Lejeune agreed to become the Director of Medical Research for the future United States - based IFGR/MF that was formally incorporated in Pittsburgh, Pennsylvania one year later.

The new foundation's International Advisory Board featured other prominent professionals including Sir William Liley of New Zealand, Drs. Marie Rethore and Marie Peeters, co-workers with Lejeune in Paris, Dr. Sigfried Ernst of West Germany, and Dr. Herbert Ratner of Oak Park IL and Professor Charles Rice of the University of Notre Dame Law School.

Michael Still an Inspiration

As most of you know, "Michael" in "The Michael Fund" is a real boy – Michael Policastro of Murrysville, PA, who was born on February 15, 1969 with Down syndrome. He is the son of Thomas and Patricia Policastro who helped create the Michael Fund.

Michael received his pre-school education at St. Peter's Development Center in Pittsburgh and his elementary and secondary schooling at Clelian Heights for Exceptional Children in Greensburg.

Michael is now 34 years old. Ask him how life is going and he'll tell you "Wonderful!" He is currently employed at Christopher's Bakery in Murrysville. Michael is a senior resident at Clelian Heights where he has lent his talents to the Gourmet Dog Biscuits program. We hope to do a full interview with Michael this coming year.

Michael Fund Launches Special Campaign to Save Human Embryos

In addition to its Down syndrome research program, in 1994-1995, the Michael Fund launched the first organized battle in the nation against non-therapeutic human embryo research conducted by the National Institutes of Health and other governmental agencies and private commercial and foundation interests in the United States.

An immediate issue was the nineteen-member NIH Human Embryo Panel that had been established in the fall of 1973 to create national guidelines for funding non-therapeutic research on human embryos. Attorneys for the Michael Fund charged that the panel was, by its own admission, stacked in favor of such research in violation of the Federal Advisory Committee Act, and that human embryo research involved the destruction of thousands of innocent human beings at the earliest stages of development.

Money for the Michael Fund lawsuit against the NIH Human Embryo Panel was raised in a separate fund-raising effort specifically designated for the Human Embryo Defense project and was not taken from our research program. Thanks to Maryland attorney Martin Palmer and a special effort by Dr. Samuel Nigro head of the Cleveland Catholic Physicians Guild and regular supporters of the Michael Fund we were able to bring our law suit all the way up to the U.S. Supreme Court which regretfully refused to take up the case.

Defenders of human embryo research have stated that the killing of these tiniest of human beings is necessary to advance genetic research, but the Michael Fund has from its beginning always rejected the 'health by death' ethic.

Dr. Lejeune harbored a special concern and love for the human embryo whom he called 'the least of our kin.'

On August 10, 1989, he testified before the Circuit Court for Blount County in Maryville Tennessee in the now famous Davis v Davis Case (No. E-14496) that involved the disposition and custody of seven cryogenically frozen human embryos produced by in *vitro* fertilization and living in concentration cans at the Fertility Center of East Tennessee.

Dr. Lejeune presented scientific evidence for the humanity of the frozen embryos and argued that they were not property that could be bartered or disposed of like inanimate objects. The judge agreed with Dr. Lejeune and awarded custody to the mother of the children, although a higher court later overturned that decision.

In a conversation with Mrs. Engel a week before his death on Easter Sunday, April 3, 1994, Dr. Lejeune expressed his hope that the battle to defend the human embryo would go on. And it has.

The Michael Fund Comes Home

After the death of Dr. Lejeune and the breakup of his

research team at the University of Paris, the Board of Directors voted to bring the Michael Fund research program home to the United States.

Dr. Marie Peeters-Ney of Nova Scotia, Canada who had worked with Dr. Lejeune in France was appointed the new Director of Medical Research for the Michael Fund. She is a specialist in the effects of folic acid, vitamins and amino acids on children with Trisomy 21.

In late 1996, Dr. Peter Kummant, Chairman of the Board announced the foundation's first U.S. grant for curative genetic research to Dr. Paddy Jim Baggot, Director of Maternal fetal Medicine and Human Genetics at the Paul VI Institute, Omaha, Nebraska.

The \$165,000 research grant project, *Biochemical Analysis of Folate-Dependent Metabolites in Amniotic Fluid*, incorporated some of the earlier research findings of Dr. Lejeune. Dr. Baggot's research centered on the biochemical deficiencies known to be relevant to Down syndrome including metabolites of organic acids, amino acids, neurotransmitters and carbohydrates.

As this newsletter goes to print, Dr. Baggot is completing his final report to the Michael Fund. These findings will then be submitted to medical journals in the United States for publication. Dr. Baggot has also finished a worldwide scientific bibliography on Down syndrome research that the Michael Fund hopes to publish at a future date.

Henry Foster Controversy

Unlike many advocacy groups for the handicapped, the Michael Fund has never had extra funds to pour into a national public relations campaign. Money has always just been too tight. Whatever local, state and national publicity we have gotten has generally come from pro-life groups and Down syndrome organizations and by word of mouth from one household to another.

In mid-February 1995 however, the International Foundation for Genetic Research/Michael Fund made national headlines in the secular press when it identified Henry W. Foster, President Clinton's nominee for Surgeon General of the United States as a skilled late-term abortionist specializing in the killing of genetically handicapped unborn children.

Government transcripts from the Michael Fund's extensive archives on U.S. eugenicists, documented Foster's commitment to eugenic abortion including the killing of suspected sick-cell anemia children in the womb. Initially both the White House and Foster labeled the NIH transcripts a forgery or mistake. But the transcript was for real and virtually overnight the nomination was dead in the water. Shortly thereafter, the President removed Foster's name from consideration as Surgeon General.

The Foster controversy marked the very first time that the Michael Fund had received attention in the national press. With that publicity the foundation was able to reach out to thousands of Americans who prefer that their

charity dollars go toward healing not killing of children with birth defects.

Michael Fund Newsletter Tops With Donors

One of the most important features of our foundation over the last twenty-five years has been the **Friends of the Michael Fund Newsletter**. Unlike the typical organization newsletter, ours has featured original articles related to historical and contemporary information on the genetics and eugenics.

The fall 1998 issue on “The Geranium in the Window” – the story of the *Kinderhaus* murders in Nazi Germany that were carried out against handicapped children at the famed Egfling-Haar Psychiatric Institute outside Munich, was probably one of the best received newsletters the Michael Fund has ever published.

The fall 2002 newsletter on “BABI: Another Anti-Life Genetic Baby Package” that exposed the horrors of pre-implantation diagnosis was also among the most informative and thought-provoking articles we have published.

Copies of these and other back IFGR/MF newsletters are available to all donors upon request.

Raising a Pro-life Genetic War Chest

Financially speaking, when the Michael Fund was created in 1979, it had to start from ground zero.

Luckily, we were blessed by a start-up grant from Harry John of the DeRance Foundation of Milwaukee, WI. that helped us with the legal costs of setting up a charitable tax-exempt foundation.

Later we also received a series of grants from the Human Life Foundation of NYC, James McFadden, Chairman. Last year we received a generous grant from the Laboratory for Education and Research in Neuroscience, Detroit, MI. that enable us to close the accounts on the Baggot research grant.

However, the largest portion of the Michael Fund budget over the last twenty-five years has come from individual donors like you.

The first Michael-Fund Walk-a-thon was held in 1980 in Long Island, NY. It was organized by veteran pro-lifer Dolores Andreski, who later became our National Fund Raising Chairman. John (Jack) Short, Associate publisher of *Down Syndrome Today* and Charlie Williamson of the Knights of Columbus were also instrumental in developing support for the Michael Fund on L.I.

One of the Michael Fund’s most memorable fundraisers was the 1982 Jeff Steinberg Concert held at Duquesne University in Pittsburgh. Jeff, known as the “Ambassador of the Handicapped” was born without hands or arms and with two malformed legs. He is one giant of an entertainer and his performance was one that the audience will remember for a lifetime.

The well-know rock band Three Way Street based in Philadelphia has also made the Michael Fund the recipient of profits taken in from its record sales.

“Actually, there shouldn’t be any controversy when it comes to the Michael Fund’s positive approach to the prevention of birth defects,” says Brian McCafferty, a consultant to Three Way Street.

Individual chapters of the Knights of Columbus throughout the United States have also supported the research program of the Michael Fund.

We would like to especially recognize the Father James J. Kelly Council, No. 3632 of the K of C of Verona-Cedar Grove-West Orange New Jersey, Stanley W. Butler and John A. Sommer, Jr., Chairmen. For many years the Fr. Kelly Council has supported the Michael Fund from funds raised by its annual K of C fund drive to benefit mentally and physically handicapped children. In 1985, the Michael Fund was also selected by the Knights of Columbus of Illinois as one of the beneficiaries of its Walk for Life statewide project to aid the handicapped child – born and unborn.

We also owe a great debt to the many physicians of the National Catholic Physician’s Guilds throughout the United States who have helped the Michael Fund in times of special financial need.

Parents of children born with Down syndrome have been among the Michael Fund’s most enthusiastic financial supporters and many have served on our National Advisory Board over the years. A special thanks to Jessie Bennett founder of Down Syndrome International, Mr. and Mrs. Donald Buell, and Drs. Richard and Leonie Watson for their inspiring assistance from day one of the founding of the Michael Fund.

The Michael Fund would also like to recognize those donors who have designated our agency (#185) as their charity of choice under the donor-option program of the United Way-Pittsburgh.

♡ Memorial Gifts

Dr. Herbert Ratner by Michael Fund Board
Thomas Policastro by Michael Fund Board
Kathy Bates by Michael Fund Board
John Short by Michael Fund Board
Robert Joseph Hammer by Lucille Hammer
Patricia Ann Peduto by Anita Peduto
Brian Cox by Mary Althoff
Mary Francesca Chada by Family and Friends
Baby Scarlett by M/M Robert Scarlett and Family
Sheila Dundon by Patrick Dundon and Friends
Dr. Joseph Stanton by Mary Stanton
Anna May Knieser by Family and Friends
Norma Marbellce by Dorothy Molder
K. J. Gemmill-Kennedy by Annette Ravinsky-Fleeger
Joseph Doyle by Mavis A. Walters
Steven A. Antol by James Grab Family
Brian and Colleen Hughes by Ray and Vivienne Kell
Lillian Zaquersky by Our Lady of Mercy, R&AS
Matthew T. Mackintosh by Family and Friends
Lynn Rydingsvard by Margaret Schrader
Jamie M. Foutz by James R. Foutz
All Unborn Children - March for Life Bus, Pgh.

MICHAEL FUND DONOR HONOR ROLL

James Rupprecht	M/M James O'Grady	M/M Harry Lydic	Rosalie Horner
Virginia Brill	Nancy Baric	Mrs. Harold House	Mike Sutkovich
Tim O'Callaghan	Albert Wigchert	Dr. James Thompson	M/M John Finn
David G. Hart	Barbara Gallagher	Elsie Brophy	Virginia Evers
Robert F. Nolan	Mary McDermott	Babara Grimaldi	Margaret Schneider
Ernest & Nita Gero	Bob Howard	M/M Joseph Kenny	M/M William Powderly
Mary Brill	Grace De Vos	Frank Spahitz	Donald Turecek
Pinellas County RTL	Joan Higly	Joseph Angelo	M/M William Galles
M/M James Keegan	Larry Pennypacker	Dr/M John Wilke	Jerry & Amy Benitz
Herman Wong	Robert Rausch	Mary C. Scanlon	Pregnancy Center, Inc.
Helen Ofsharick	Eileen Egan	Alice Drennan	Harry Failla
Effingham RTL	St. Thomas RTL	Linda Formanek	Anthony Cristofich
Maureen Zack	M/M Joseph Jordan	Joseph Wall	James Winker
M/M Joseph Bartish	William Forgacs	Joseph Warganz	Marie Feeley
Diocese Rockville Ctr.	Dr. John Hackett	Russell Pond	M/M Robert Vogel
John Day	Lucille Koehler	NH ProLife Council	Danielle Limberg
Mary & Paul Althoff	Mary Veres	Mary O'Hara	George Bertoloti
Lumen Christi TORCH	M/M Richard Conklin	Joann T. Dreist	Vaughn Family
Crusade for Life	Michael Pennacchia	Chales Van Hecke	John Rosser
Mr. Mrs. John Reuter	M/M Gary Zack	M/M Jeffrey Finck	Joanne Slappo
Anita Peduto	Jolane Ceccni	M/M Joseph Esker	Doug Faber
Zora & John Darrow	David Thiel	Mrs. Cleta Jasper	Norbert Romie
Betty Hartzell	Terri Vandcoevering	James Hearn	Dan & Annette Osen
Henry County RTL	M/M Paul Tobin	M/M Dan Schoenecker	Dana Slattery
Marie E. Feeley	M/M Robert Duggan	Charles Dawson	Patricia Bergheiser
James Winker	Helene Burgman	M/M Jack Koester	Harriet Davison
M/M Lou Eskuchen	Juanita Mattingly	Peter Moody, Jr.	M/M Robert Rausch
Charles Foy	M/M Robert Hansen	M/M Gerard Weigel	M/M James Feltz
Erik & Cathy Rivers	M/M Steve Blubaugh	R.W. Hermann	Tom & Kathy Benton
M/M M. Cassidy	Gary Lucas	Charles Gilligan	Huseman-Donovan
Patrick Dundon	Thomas Youket	John and Olive Linn	Family
Fr. Mike Chapman	Marvin Wegman	Rev. George Limmer	Goldie Mischel
Dorothy Gillen	Ann Trojan	Kate Adams	Susan Gruber
Mr. Robert Scarlett	Richard Gorsline	Dr/M John Gillette	Marian Smyth
William Sharrar	Michael Criello	Kathleen Barrett	Mrs. Sybil Paffrath
Wagner RTL	M/M Frank Kelly	Joan Varacalli	Mary Meehan
Conrad Beaulieu	Betty Cummings	M/M Jeffrey Finch	Margie Mineweaser
Vivienne Kell	Joseph M. Yates	Veronica Devlin	Richard Steinmetz
John T. Walsh	M/M Thomas Niblo	Joseph Bendel	Charles & Agnes George
Rev. C. Hill and Family	M/M John Cunningham	Regis Marinchek	Mike Sutkovich
N.J. Committee for Life	Tom Henn	Rita Burns	M/M Paul Keteles
St. Anthony's Church	Jenny Biondi	Naussau-Soffolk	Dan Piroutek
Rev. John P. Moloney	Joan Higby	Knights of Columbus	Jean Seidl
Gale Folsland	RTL Coalition of	Dr/M Robert Martin	David & Kathy Brown
Lucille Hammer	Lower Westchester	Frank Grabarits	Richard Onken
Bascom LIFE	Dr. Philip Dreisback	Patricia Hayes	Therese M. Torres
St. Linus Pro-Life	and Family	Joan Mullane	M/M James Korff
Catholic Daughters of	M/M Norman O'Grady	Bridgid Kernan	Duane Ruud
America	John Stasa	Anthony Coomes	Frances Di Persia
People Concerned for	M/M Robert O'Neill	Gerry Parmantier	Raymond Merisko
the Unborn Child	Pauline Murphy	Dr. John Dougeveto	Rosemarie Simon
Virgina King	M/M George Pavlic	Ann Keen	M/M Robert Siman
Fr. A. Shaughnessy	Dr/M George Deitz	Joan Solms	M/M James Massura
Sheila Wharan	Barbara Syska	Jeannette Wansing	Katherine Kuester
James F. Brennan	M/M Lawrence Smith	M/M Charles Overbeck	(To be continued)

Leland Richardson
Victoria Sferlazza
Loretta De Ponio
(*To be continued)

is ultrasound. It can be used for good or ill. It is inexpensive and is generally considered safe. Ultrasound can detect anatomic or structural birth defects including spina-bifida, cardiac anomalies, abdominal defects and various forms of urinary obstruction. Despite the fact that these can be treated surgically after birth, abortion of affected children may still be offered as a "choice."

For chromosomal abnormalities and other genetic disorders amniocentesis is used. The procedure is carried out from 16-20 weeks gestation and involves placing a needle into the fluid-filled amniotic cavity surrounding the baby. Fluid is withdrawn for diagnostic testing. The risk of miscarriage from the procedure is about 1/200.

Chorionic villus sampling (CVS) samples the placenta and is carried out earlier than amniocentesis, usually at 10-14 weeks. Limb reduction anomalies have been linked to CVS and the risk of procedure-related

miscarriage is estimated to be 1-2%.

Fetoscopy or percutaneous umbilical blood sampling (PUBS) involves the use of ultrasound guidance to obtain a blood sample from the fetal umbilical vein. Fetal death from the procedure is 2-6%.

Mrs. Engel: *Are there any therapeutic or life-saving uses for these procedures?*

Dr. Baggot: Yes, for amniocentesis. But here the timing and intent of the procedures are different.

For example, some amniocenteses are done after 32 weeks to determine if the fetus' lungs are mature enough for delivery. Here the intent is strictly therapeutic. These do not result in miscarriage or abortion, hence there is no moral objection to these procedures.

Generally speaking, the risks are lessened by delaying amniocentesis until after viability and performing the procedure well. The benefits are increased

by including tests for treatable disorders in the fetus although currently these disorders are few in number.

Mrs. Engel: *Can you give us an example of a biochemical genetic disorder?*

Dr. Baggot: I think the archetype of biochemical genetic disorders in phenylketonuria (PKU). Children with PKU are unable to metabolize phenylalanine one of twenty amino acid building blocks of proteins. Excessive levels of phenylalanine are toxic to brain development and as a result these children previously had mental retardation.

Today these children can be treated with a phenylalaine restriction diet. If these children are detected at the time of birth and treated shortly thereafter, they can have normal intelligence.

This disorder can be detected prenatally either by measuring phenylalaine in amniotic fluid or by culturing fetal cells and assaying their ability to metabolize phenylalaine.

Mrs. Engel: *What are the various diagnostic procedures used for prenatal diagnosis?*

Dr. Baggot: The most widely used procedure

Brian Cox There are about 700-1000 biochemical disorders, most of which are very rare. About seventy of these have treatment. The overall frequency of all these disorders taken together is about 1/200.

Mrs. Engel: *What about molecular genetic disorders?*

Dr. Baggot: Molecular genetic disorders are caused by misspellings in the DNA sequence of a particular gene. One such archetype of a molecular genetic disorder is cystic fibrosis (CF), which is a respiratory disease caused by a malfunction of a protein.

Cystic fibrosis was initially known as mucoviscidosis because children with this disease were unable to excrete their pancreatic enzymes into the intestines that were blocked by viscous accumulations of mucoid secretion. As a result these children died of malabsorption in the first one or two years of life. With pancreatic enzyme replacement however, their life span has been vastly increased.

Now these children develop severe chronic respiratory disease. As in the pancreas, the airways in children with cystic fibrosis are often blocked by viscous mucoid secretions which contain degenerating cells. Within these degenerating cells are nuclei and within the nuclei are chromosomes that are composed of long filaments of DNA. One reason for the high viscosity of these secretions is the presence of numerous long strings of DNA. This situation can be treated by the application of an enzyme that chews up DNA, known as DNase. In ideal climates where lung infection is minimized, some patients with cystic fibrosis survive past the age of fifty.

Mrs. Engel: *In recent years the media has given a great deal of attention to mass screening for CF has it not?*

Dr. Baggot: Yes. Mass maternal screening for CF has been suggested by some. Molecular genetic techniques can be used to determine if the mother is a carrier. If the mother is screened positive then the father can be tested. If both mother and father are carriers, then an amniocentesis followed by the abortion of an affected child is frequently recommended. Such screening programs are in actuality eugenic programs designed to reduce or eliminate the birth of babies with CF.

We can see, using the CF model, that the rapid strides being made in the treatment of cystic fibrosis have not yet eliminated the desire to kill the unborn child afflicted with the disorder. This eugenic mindset is unfortunate because we know that when today's adults with CF were born, their prognosis was dismal, whereas children born today given the scientific progress in treating the disorder, are doing much better than expected when they were born. And I think that it is reasonable to hope that this progress will continue into the future.

So I think it is reasonable to encourage a mother to refuse an abortion, carry her baby to term even though she has been advised that the child has CF, and hope that things will turn out better than expected. What cliché is more worn out than the baby with a disease who does better than he/she was expected to do?

Mrs. Engel: *Thus far we have talked about genetic disorders from the perspective of an affected unborn child. What about a pregnancy where the mother has a genetic disorder and a "maternally-indicated" abortion is advocated to "save her life"?*

Dr. Baggot: Let us look at the case of Marfan syndrome which is alleged to have a high mortality in pregnancy and for which a "therapeutic" abortion is almost always recommended to preserve the life of mother with the disorder.

Mrs. Engel: Can you first describe Marfan syndrome and how the disease manifests itself?

Dr. Baggot: Marfan syndrome is inherited in an autosomal dominant fashion. This means that the disorder results when a single mutation on either the maternal or

paternal copy of the gene. In the case of Marfan syndrome the mutation is in the fibrillin gene on chromosome 15.

Marfan syndrome is characterized by tall stature, skeletal abnormalities, dislocation of the ocular lens and cystic medial necrosis of the arteries. The latter causes aortic dissection and rupture, and cardiac valve abnormalities. Persons thought to have Marfan syndrome need to be carefully diagnosed as it can be confused with Stickler syndrome, which is much more common, or with another disorder called homocystinuria.

Mrs. Engel: Does pregnancy worsen Marfan syndrome?

Dr. Baggot: No. Adult maternal patients with Marfan syndrome have a shortened life span and a high mortality, regardless of pregnancy. Recent studies have shown that the cardiovascular outcome in such patients was not different from similar Marfan patients who were not pregnant.

Mrs. Engel: *Why then are mothers with Marfan syndrome told that an abortion is "medically-indicated"?*

Dr. Baggot: Why indeed?

First, I think that most of your readers will appreciate the fact that in the case of Marfan syndrome the problem is with the cardio-vascular system not the reproductive system. An abortion kills the patient's preborn child (who may or may not have the disorder) but does nothing to treat the underlying vascular disease nor prolong the patient's life. Therefore from the maternal perspective abortion is not indicated to either "save" or "improve" maternal outcome.

Second, many physicians base their advice to abort on older literature on Marfan syndrome that was published before the remarkable era of cardiac medical and surgical therapy. I think this factor holds true for many diseases for which an abortion is said to be "maternally-indicated," although that list is shrinking dramatically given certain advances in medical science.

Third, the management of serious maternal disorders in pregnancy often involves interdisciplinary situations. Physicians are often comfortable with all the risks of their own specialty, because they have abundant experience and know alternative remedies for each problem. For situations outside their specialty, they may be uncomfortable due to lack of knowledge or experience.

Lastly, I think there is the human factor. Physicians as a group tend to be cautious in their evaluation of patient outcomes. They are also highly cognizant of the possibility of a lawsuit being filed against them should their patient die – so they recommend an abortion. I believe that it is unfair to advise abortion for fear of lawsuits, without disclosing the conflict of interest to the patient.

Mrs. Engel: *What if the Marfan patient becomes pregnant again?*

Dr. Baggot: Well, from the mother's perspective she has not gotten any younger and she still has her heart problem – so essentially her situation has not changed. As for the physician he may recommend another abortion or, if he has gained in wisdom, he may refer the mother to a cardiac specialist who can truly help the mother without harming the child.

Mrs. Engel: *Is there a built-in bias in the field of genetics that favors eugenic abortion over treatment of the affected child in the womb?*

Dr. Baggot: Yes, I think there is a bias that genetic disorders are untreatable. This idea stems from the fact that when the current generation of physicians were in medical school, the adjective "genetic" strongly suggests that a disorder was untreatable. For most of us it is breathtaking that each month new treatments are being reported for genetic diseases.

Mrs. Engel: *Would you highlight for us some of these new treatments?*

Dr. Baggot: Earlier, we have discussed the disease phenylketonuria (PKU) which is a biochemical disorder that can be treated by restriction of the substrate in the diet.

There are other biochemical disorders that involve the production of a defective protein. Frequently the defective protein is an enzyme. In many cases the defect of the protein derives from the fact that it only binds weakly to co-factors which are essential for its proper function. Provision of what would normally be seen as excessive amounts of the co-factor can overdrive a weak enzyme to the point that its function is more normal. One example would be the treatment of maple syrup urine disease with thiamine (vitamin B1). This category of treatment represents protein activation with co-factors.

Then there are the skeletal disorders, which include a large number of genetic disorders. These can be treated today by standard clinical means and these therapies are rapidly improving. Thus surgical reconstruction for skeletal disorders is an example of clinical treatment.

There are various types of congenital adrenal hyperplasia in which there is a weakness in the ability to synthesize essential products. In this category of disorders the treatment represents product replacement.

Another category of treatment is protein replacement. In this type of treatment a protein product which is not being produced in the proper amounts is replaced as in the treatment of hemophilia A with factor VIII.

There is also transplantation therapy as in the case of a bone marrow transplant for beta-thalassemia.

Thus, even excluding the prospect of new gene therapies that loom on the horizon, we already have many treatments already available for genetic disorders.

Mrs. Engel: *What about the field of fetal therapy in obstetrics?*

Dr. Baggot: This is an area in which cases need to be very carefully individualized.

As a rule, pediatric surgeons of different surgical specialties prefer to have their patients in the best possible condition for surgery. As long as the patient is not getting worse, his or her ability to withstand and recover from surgery may improve with advancing gestational age.

This consideration is often an impediment to fetal surgery where the risk of premature delivery and fetal death exercises a formidable difficulty. Leaving a scar on the wall of the uterus is a significant irritant to the womb and uterine irritability means contractions.

Therefore, in many cases, post-natal therapy is often better than intrauterine surgery especially in the case of anatomic birth defects. Thus children with hydrocephalus can be shunted postnatally. Babies with spina bifida can be repaired after a cesarean delivery. Some urinary tract obstructions can also be treated postnatally. In such cases the risk of inducing premature birth and fetal death outweigh the advantages of intrauterine fetal surgery.

Mrs. Engel: *Are there cases in which such intervention would be justified?*

Dr. Baggot: Yes, there is the opposite side of the coin, in which the pre-born child's condition is serious and getting worse, and intervention would truly be remedial. The fact however, that fetal surgery is so rare in part reflects that the latter consideration does not often apply.

Mrs. Engel: *Birth defects affect approximately 3-5% of all children at the time of birth. Would you like to say a word about the causes and prevention of birth defects?*

Dr. Baggot: There are many causes of birth defects, both multiple and isolated.

They include infections, such as rubella, syphilis, and cytomegalovirus. Also, drugs of abuse such as cocaine are frequent causes of birth defects, as are the more common use of alcohol and tobacco.

There are certain commonly used medications that may cause birth defects, especially anti-seizure medications.

Certain disorders such as diabetes and phenylketonuria can cause birth defects, which is why it is important to control these disorders before conception.

We know that environmental toxins can lead to birth defects but it is difficult to know how common they are as causes of birth defects.

Of course we have known the link between nutritional deficiencies and excesses and birth defects for a long time.

Mrs. Engel: *This brings us to the realm of the prevention of birth defects before birth.*

Dr. Baggot: As a prolife geneticist-obstetrician, I am always happy to address this issue.

I think it is laudatory to want to prevent birth defects, but I reject the idea that eugenic abortion "prevents" birth defects. It does not. It's just abortion.

In cases where the causes of birth defects are related to specific human behaviors such as illicit drug, the "prevention" of birth defects is, I think, self-explanatory.

In terms of nutritional deficiencies (and excesses) I think this is an area of great promise in terms of preventing birth defects. For example, there are the well-publicized benefits of folic acid in preventing neural tube defects such as spina bifida and anencephaly. What is less well known is the fact that it has also been shown to prevent urinary tract anomalies, some cardiovascular defects, some limb deficiencies and cleft lip and cleft palate.

There are also birth defects caused by either a deficiency or excess of certain vitamins including vitamins A, B, D, and E as well as certain minerals such as calcium, magnesium, potassium, copper, iodine, manganese, zinc, selenium and chromium.

Mrs. Engel: Thank you Dr. Baggot for all your insights – both medical and philosophical.

Dr. Baggot: And I thank you and all the supporters of The Michael Fund who have made my grant for Down syndrome research possible.

References: Beaudet, A.L., Scriver, C.R., Sly, W.S. & Balle, D. *The Metabolic Basis of Inherited Disease*, McGraw-Hill, Inc., NY, 1995.

Rossiter, J.P., Repke, J.T., Morales, A.J., Murphy, E.A., Pyeritz, R.E., "A Prospective Longitudinal Evaluation of Pregnancy in the Marfan syndrome," *Am J Obstet Gynecol.*, Nov. 1973 (5): 1599-1606

More About The Michael Fund

The International Foundation for Genetic Research, popularly known as The Michael Fund, was created in 1978 as a prolife alternative to the March of Dimes. All contributions and memorial gifts are tax-deductible. For -

- Back issues of our newsletter
- Information on our Down syndrome research program
- Physician referrals for specific birth anomalies
- Information on adoption of handicapped children including children with Down syndrome
- Breast-feeding and dietary management of children with Down syndrome

Contact us at www.michaelfund.org
or call 412 – 823 - 6380



