

Geno - Terato Epi - Tome



Editor: W. Wertelecki, MD

July 2006

Please circulate. Print additional copies from <http://www.ibis-birthdefects.org/start/pdf/200607GenoTerato.pdf>

“Every Child Succeeds” Saves Infant Lives A Proctor & Gamble Company Approach

In the fight against high infant mortality rates, Cincinnati took a novel approach and applied a corporate model. The results were remarkable. In Cincinnati, 13 out of every 1,000 newborns die by the age of one year. This program, called “Every Child Succeeds” (ECS), reduced the rate to 2.8 (the 2004 US average is 6.8).

ECS is designed to improve maternal and infant health and is built on a tough corporate model inspired by a former chief of Proctor and Gamble Co., (three retired P&G executives volunteer their services). ECS targets pregnant women and new mothers who are poor and usually single, and helps mothers from pregnancy until the child’s third birthday. A team of social workers and nurses visit new moms in their homes and “hector” them to eat better, stop smoking, and breastfeed their baby. Over a period of three years, roughly 75 home visits are made at an annual cost of about \$2,500 per family. Various tools from the corporate world are used.

Details in: <http://www.everychildsucceeds.org/>

Comment: A highly sensible approach – a home visitation program. A similar strategy was used by PCAP (Parent Child Assistance Program – a paraprofessional home visitation model for extremely high risk substance abusing women). Both programs were effective. In Alabama, the Alabama Fetal Alcohol Spectrum Disorder Prevention Initiative (AFI) and state programs to reduce infant mortality should consider adopting the strategies used by ECS and PCAP programs.

For information about AFI, please visit http://www.southalabama.edu/genetics/bdsp_fasd_index.htm or contact bo-liver@usouthal.edu

Childbirth Risks amid Soaring Liability Costs

Laura Landro in the Wall Street Journal of July 12, 2006, underscores that at the root of 85% of all adverse events reported in the obstetric units are communication breakdowns. Five other top contributors to obstetrics litigation are failures to recognize fetal distress, timely Caesarean birth, proper resuscitation of depressed babies, inappropriate use of labor – inducing drugs, and vacuum/forceps. Some of the more-or-less inflammatory comments in the article include “Pitocin is used like candy in the OB world ... it is common practice “to pit to distress” – or use the maximum dose of pitocin to stimulate contractions ... obstetricians are not used to having their wings clipped ... it is hard to get doctors to go along because they don’t necessarily believe the risks ... research shows that delivering babies even a few days early is associated with higher rates of emergency Caesarean deliveries, and admissions to the neonatal intensive care unit ... the obstetricians don’t always adhere to guidelines for elective induction .. they often schedule deliveries around their own office hours or travel plans ...”

Details: infomedpatient@wsj.com

Down Syndrome – Major Scientific New Vistas Deleting Extra Genes may Help

“You can stop thinking of Down Syndrome as this murky area where you will never discover specific genes that make a difference ... if we can decrease the expression of this gene, we may be able to provide something more than supportive care for people with Down Syndrome ... we may now have an opportunity to make a big difference in people’s lives”, said neurologist William Mobley, MD, PhD, who is the John E. Cahill Family Professor at Stanford’s School of Medicine and Director of Lucille Packard Children’s Hospital’s Down Syndrome Center.



It is estimated that nearly 300,000 persons in the United States have Down Syndrome. Mobley’s team focused its attention on the gene for amyloid precursor protein (App) found in Alzheimer disease. The studies were conducted on a trisomic mouse model of Down Syndrome (with three App genes). The investigators showed that, when the extra copy of the App gene was deleted, the otherwise trisomic mice, developed “more normal”. The investigators also showed that neuronal cell death of trisomic mice could be prevented by deleting another extra gene (TrkB).

Details in: June 2006 issue of the Journal Neuron and in <http://www.the-scientist.com/news/>

Comment: The findings are tantalizing and have called the attention of the world press. The Alabama Birth Defects Surveillance and Prevention Program offers information to medical homes and families of infants detected to have Down Syndrome (Trisomy 21). Early care and multi-disciplinary follow-up are critical to optimize development of children with Down Syndrome. The prevalence rate of Down Syndrome in Alabama is not significantly different from other states (~ 12 per 10,000 live births).

ACE Inhibitors Cause Major Congenital Malformations

W. O. Cooper, et. al. studied nearly 30,000 infants enrolled in the Tennessee Medicaid system, 209 of whom were prenatally exposed to ACE Inhibitors. Prenatal exposure to ACE Inhibitors was associated with an excess of major congenital malformations, the risk being 2.7 times greater than for those not exposed. Atrial septal defects in combination with other cardiac malformations were most common. Also common were spina bifida, microcephaly, coloboma, renal and intestinal dysplasia, as well as diaphragmatic hernia.

Details in: William O. Cooper, et.al, “Major Congenital Malformations after First-Trimester Exposure to ACE Inhibitors”, NEJM, 354:23, June 6, 2006.

Comment: Over 50% of pregnancies are not planned and women of reproductive age should avoid exposing the unborn to ACE Inhibitors, a significant challenge in view that the use of such agents has rapidly expanded.

Contact us: Geno-Terato-Epi-Tome, Department of Medical Genetics, University of South Alabama
Email: wwertele@usouthal.edu

About clinical services, genetic laboratory testing, counseling and paternity testing
Email: shudson@usouthal.edu or Call Toll Free: 1-800-624-1865

Websites:

- Alabama Birth Defects Surveillance and Prevention (<http://www.usouthal.edu/genetics/>)
- International Birth Defects Information Systems (<http://www.ibis-birthdefects.org/>)
- Medical Humanities (<http://www.pandorawordbox.com/>)