

Presentation 2 – Betty Mekdeci



BIRTH DEFECT
RESEARCH FOR CHILDREN



National Birth Defect Registry

- Based on concept of “alert practitioner”
- Identifies patterns of birth defects with the same or similar exposures
- Collects data on maternal/paternal pre-conceptual and prenatal exposures
- Cases dually identified by individual symptoms and overall syndrome or category name
- Designed to raise hypotheses

**Teratogens Identified
by Alert Practitioners**

- Thalidomide
- Rubella
- Dilantin
- DES
- Alcohol
- Methyl Mercury
- Radiation

Scientific Advisory Board

- | | |
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BIRTH DEFECT RESEARCH FOR CHILDREN, INC.
REGISTRY QUESTIONNAIRE
(Please print in blue or black ink or no.2 pencil)

Date: / / Reporting person's name: _____
Relationship to child: mother stepmother grandmother other: _____
 father stepfather grandfather

Street/POB: _____ Apt No: _____
City: _____ State: _____ Zip: _____
Phone: (____) _____

Name and number of someone who can always get in touch with you: Name: _____ Phone: _____

I would like to be matched with other families of children with similar disabilities. yes no

CHILD DISABILITIES INFORMATION

Child's name: _____
Sex: Male Female Blood Type: A B AB Rh+ Rh- Race: Asian or Pacific Islander Native American or Alaskan Native Black (non-Hispanic) White (non-Hispanic) Hispanic Other (please specify): _____

Date of birth: / / Place of birth: Child's age at birth (months of gestation): _____
Birth weight: lbs. oz.

Do the father and mother of this child have other biological children? yes no don't know
Do not have birth defects? yes no don't know

If yes, please list date of birth and sex for each one:

DCB: <u> </u> / <u> </u> / <u> </u> <input type="radio"/> Male <input type="radio"/> Female	DCB: <u> </u> / <u> </u> / <u> </u> <input type="radio"/> Male <input type="radio"/> Female
DCB: <u> </u> / <u> </u> / <u> </u> <input type="radio"/> Male <input type="radio"/> Female	DCB: <u> </u> / <u> </u> / <u> </u> <input type="radio"/> Male <input type="radio"/> Female

Were child's disabilities diagnosed by a: physician teacher other: _____
 psychologist social worker

Are medical or school records available confirming child's disabilities? yes no don't know
Would you be willing to be contacted for further research? yes no don't know
Has your child died? yes no If yes, list cause: _____ and Date: / /

CHILDHOOD DISABILITIES
Please fill in the bubble by each condition your child has. If your child has a condition that is not listed, write the name of the disability in the OTHER space in the appropriate column.

<input type="checkbox"/> AD01 anencephaly	<input type="checkbox"/> AD10 spina bifida occulta
<input type="checkbox"/> AD02 microcephaly	<input type="checkbox"/> AD11 holoprosencephaly
<input type="checkbox"/> AD03 hydrocephaly	<input type="checkbox"/> AD12 epina bifida
<input type="checkbox"/> AD04 polycephaly	<input type="checkbox"/> AD13 Dandy Walker malformation
<input type="checkbox"/> AD05 Arnold Chiari malformation	<input type="checkbox"/> AD14 tubular stenosis
<input type="checkbox"/> AD06 cervical blindness	<input type="checkbox"/> AD15 ataxia
<input type="checkbox"/> AD07 encephalomalacia	<input type="checkbox"/> AD16 cerebral palsy
<input type="checkbox"/> AD08 absence corpus callosum	<input type="checkbox"/> AD17 other CNS
<input type="checkbox"/> AD09 spina bifida	

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Registry Projects

- Bendectin
- Agent Orange
- Gulf War
- Community based
 - Dickson, TN - oral cleft cluster

Dickson Cleft Palate Investigation

- 19 cases of cleft palate/2 expected
- TCE leachate from old landfill
- Toluene emissions from printing company
- Excessive Trihalomethanes in water
- CDC confirmed cluster
- EPA contracted new report on landfill
- Landfill now capped
- Tennessee legislature voted to move source of community's water to a different river

Gulf War Exposures

• DEET	• Dursban	• Malaria Pills
• Permethrin	• Dichlorvos	• Campfires
• Pyridostigmine	• Ficam	• Leishmaniasis
• Pentachlorophenol	• Carbaryl	• Chemical warfare agents
• Benzocaine sulfur	• Lindane	• CARC
• Aluminum phosphide	• Malathion	• Vaccinations
• Baygon	• Oil Well Fires	• D-phenothrin
• Boric Acid	• Leaded fuels	• Allethrin
• Sevin	• Depleted Uranium	• Paint
• Amidinohydrazone	• Solvents	• Others
• Diazinon	• Decontam agent	

Disability Frequency

• Resp. Infect.	117	• VSD	35
• Ear Infect.	92	• Drug Reaction	34
• Motor Delay	86	• Hypotonia	34
• Colic as Baby	62	• Seizures	33
• Speech	62	• Acne-like Rash	33
• Allergies	57	• Thrush	32
• Jaundice	56	• Mood Swings	32
• Reflux	52	• Pneumonia	29
• Heart Murmur	51	• Birth Mark	28
• High Fevers	46	• Stomach problems	28
• Ext. Ears	40	• Sleep disorder	26
• Asthma	38	• Walks on tiptoes	26
• Food Sens.	38	• Goldenhar	26
• Eczema	35	• Up. Limb Reduction	25

Male-Mediated Birth Defects

Evidence from Experimental and Epidemiological Studies

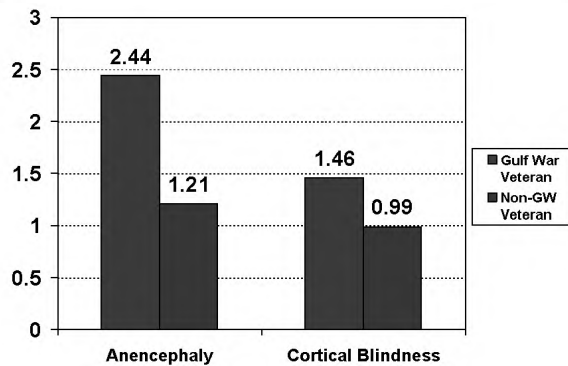
- Toluene
- Benzene
- Lead/mercury
- Trichloroethane
- Solvents
- Dyes
- Cigarette smoke/alcohol
- Drugs

Possible Outcomes

- Congenital malformations
- Spontaneous abortions
- Low birth weight
- Childhood cancers
- Developmental, neurobehavioral, neuroendocrine, neurochemical abnormalities

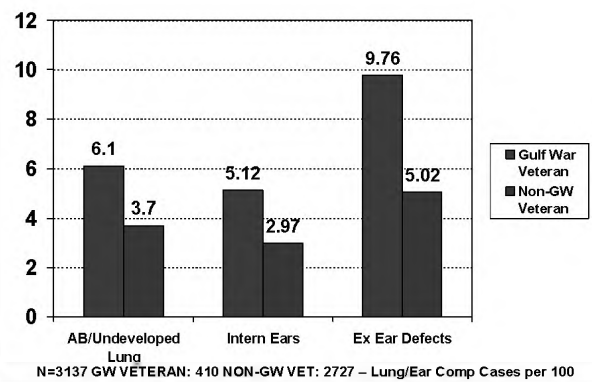
Birth Defect Research for Children, Inc.

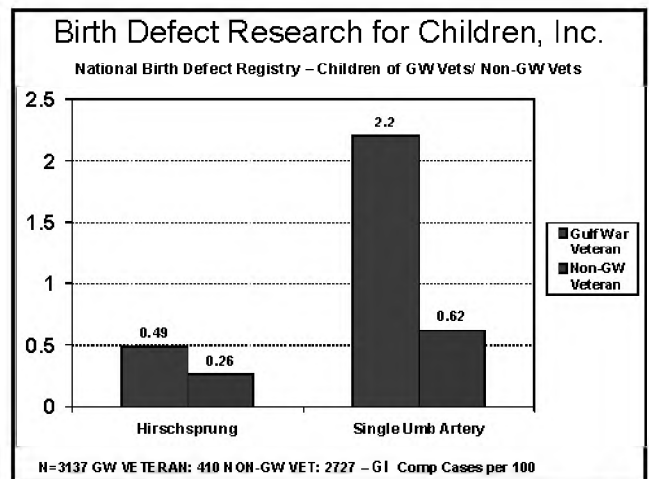
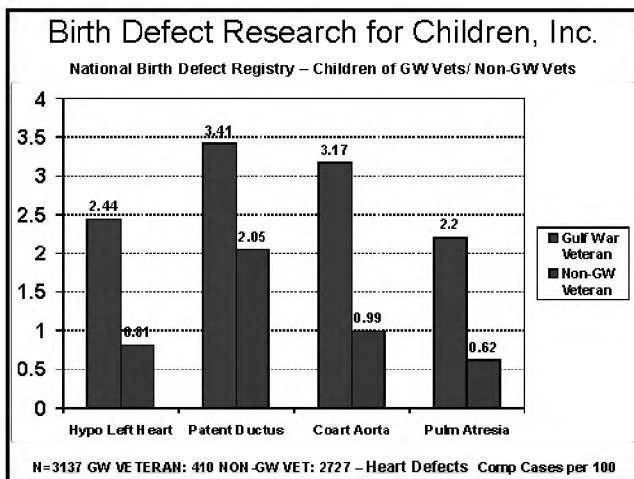
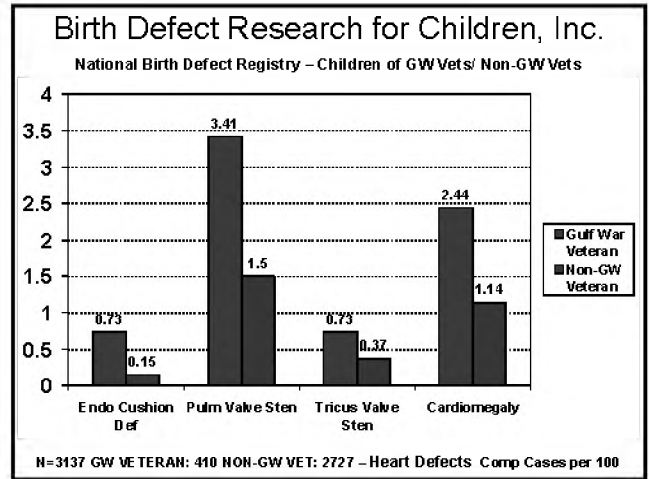
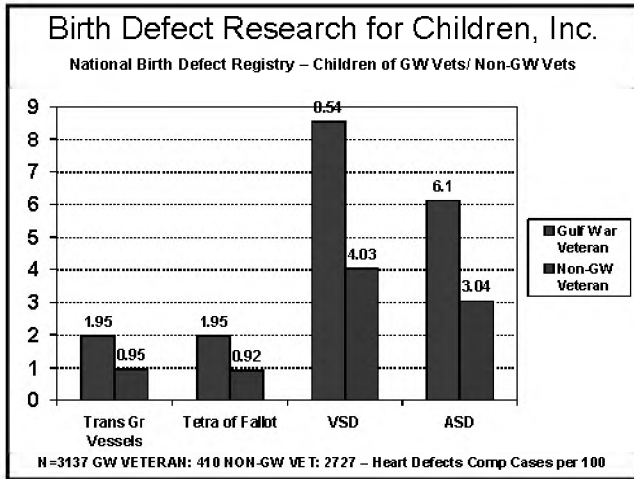
National Birth Defect Registry – Children of GW Vets/ Non-GW Vets

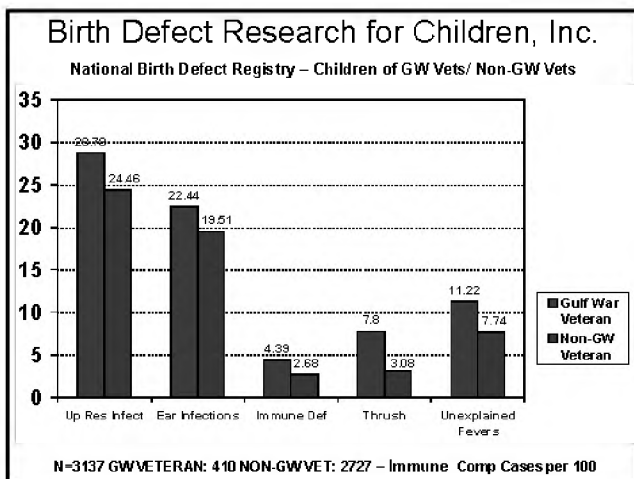
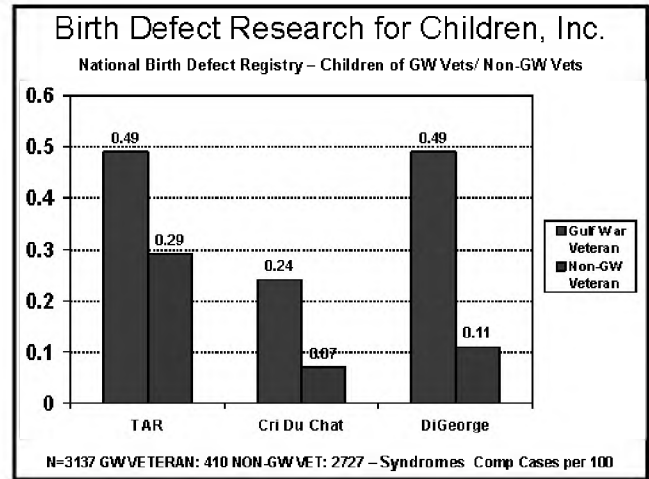
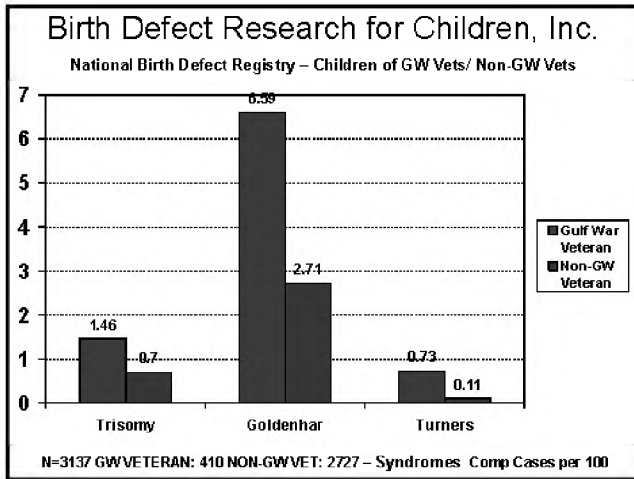


Birth Defect Research for Children, Inc.

National Birth Defect Registry – Children of GW Vets/ Non-GW Vets







Goldenhar Syndrome

- Incidence ranges from 1/14,699 to 1/45,000 births
- Associated with teratogens
 - Thalidomide
 - Primidone
 - Accutane
- Some cases familial
- Others "phenocopies"
- Aranetta study found tripling of Goldenhar in GW children born in military hospitals

Goldenhar Case Study

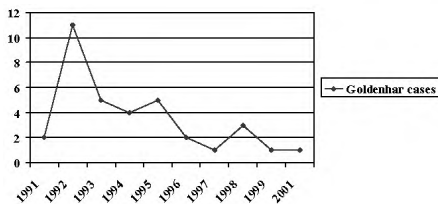
- 24 Goldenhar – fathers deployed
- 7 Goldenhar criteria fathers deployed
- 2 Goldenhar fathers in Gulf after war
- 33 Goldenhar associated with Gulf service

Goldenhar Case Study

7 Goldenhar fathers non-deployed

23344	Steel worker
22970	Shots/solvents
26815	Cont gear
26896	Shots/Naptha
30868	Shots/lead fuel
45947	mechanic
48695	Cont gear

Goldenhar Case Study DOB Distribution



Goldenhar Case Rates

- 16 D Goldenhar DOB prior 10/1/93
- 3 ND Goldenhar DOB prior 10/1/93
- 56,782 births to deployed vets*
- 68,908 births to non-deployed vets*

Crude rates for Goldenhar:
 Deployed – 28 per 100,000
 Non-deployed – 4 per 100,000

*Araneta, Teratology, 1997.

Goldenhar Details

- Medical records confirmation 14/19
- 9/16 healthy children before GW
- 2/16 healthy children after birth of child with Goldenhar Syndrome
- 16/16 Goldenhar first born after GW