How Environmental Conditions Lead to Congenital Defects

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Congenital Defects

• A congenital defect is a health problem or physical abnormality present at the time of birth.

• The severity of the defects can range from very mild, such as a slight discolor of the skin, to life threatening.
Congenital Defects

- In fact, 3.5 percent of all babies are born with some type of defect.
  - That’s 140,000 babies born in the US per year.

![Bar chart showing annual number of U.S. children born with or developing long-term medical conditions](cdc.gov)
Causes of Congenital Defects

• **Genetic factors** are associated with alterations and mutations within the genome.
  – Down Syndrome
  – Autism
  – Marfan’s Syndrome

• **Environmental factors** are usually something in the environment that the mother is exposed to during her pregnancy.
  – Growth Deficiency
  – Mental Retardation
  – Facial Defects
Genetic Factors

- **Genetic factors** are associated with alterations and mutations within the genome.
  - Gene mutations
  - Expos

http://www.youtube.com/watch?v=0wrNxCGKCws&feature=related
http://www.nasa.gov/topics/solarsystem/features/uv-exposure.html

Nasa.gov
• **Environmental factors** are usually something in the environment that the mother is exposed to during her pregnancy.
  – Lack of vitamins and malnutrition
  – Certain prescribed medications
  – Alcohol and Drugs

[Image of a pregnant woman holding a wine glass next to an illustration of a baby with a cleft palate and hare lip]

[Link: http://www.drugrehabs.ca/professional/resources/health-and-medical/fetal-alcohol-spectrum-disorder/21102010]
At the Cellular Scale

Alteration DNA

ENVIRONMENT
Influence on Signaling

GENETIC
How do scientists study these factors?

• An **animal model** is a living, non-human animal used during the research and investigation of human disease.

  ![Animal models](Mas.monash.edu)

  ![Animal models](Indiana.edu)

• The purpose is to better understand the disease without the added risk of causing harm to an actual human being during the process.
The Institutional Animal Care and Use Committee (IACUC) is a self-regulating entity that, according to U.S. federal law, must be established by institutions that use laboratory animals for research or instructional purposes to oversee and evaluate all aspects of the institution's animal care and use program.
Embryonic Development of Species

Embryo Formation

Heart Formation

Fish  Salamander  Tortoise  Chick  Hog  Calf  Rabbit  Human

Gilbert, DevBio, 8th Edition
Ex-Ovo Chick Culture System

Yalcin, An ex-ovo Chicken Embryo Culture System Suitable for Imaging and Microsurgery Applications, Jove 2010

How Scientists use these Models to Understand Congenital Defects

1. What are you interested in testing?
2. Perturb the system and observe the experiment!
Scientific Framework

• What are you interested in testing?
  – Introduction
• What do you think will happen when you test it?
  – Hypothesis
• How will you qualitatively and quantitatively record what you are observing?
  – Methods
• Record the measurements.
  – Results
• What do these results mean on the context of your experiment?
  – Discussion
Malnutrition

• A condition that occurs when your body does not get enough nutrients
Malnutrition Leads to Heart Defects

Atrial Septal Defect (ASD)
- Opening Between Atria
- Tricuspid Valve
- Pulmonary Valve
- AO = Aorta
- PA = Pulmonary Artery
- LA = Left Atrium
- RA = Right Atrium
- LV = Left Ventricle
- RV = Right Ventricle
- Oxygen-rich Blood
- Oxygen-poor Blood
- Mixed Blood

Hypoplastic Left Heart Syndrome
- Very Small Aorta
- Vessel Connecting Aorta and Pulmonary Artery
- Opening Between Atria
- Underdeveloped Left Ventricle
- AO = Aorta
- PA = Pulmonary Artery
- LA = Left Atrium
- RA = Right Atrium
- LV = Left Ventricle
- RV = Right Ventricle
- Oxygen-rich Blood
- Oxygen-poor Blood
- Mixed Blood
Retinoic Acid (RA)

Vitamin A Metabolite

### Supplement Facts

<table>
<thead>
<tr>
<th>Amount Per Serving</th>
<th>DV %</th>
<th>Amount Per Serving</th>
<th>DV %</th>
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<tbody>
<tr>
<td>Vitamin A (as Vitamin A Acetate)</td>
<td>3500IU</td>
<td>70 %</td>
<td>Copper (as Copper Citrate)</td>
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<tr>
<td>Vitamin C (as Ascorbic Acid)</td>
<td>60mg</td>
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<td>Manganese (as Manganese Citrate)</td>
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<td>Vitamin D (as Cholecalciferol)</td>
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<td>Chromium</td>
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<tr>
<td>Vitamin E (as dl-alpha Tocopheryl Acetate)</td>
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<td>Molybdenum</td>
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<td>Vitamin K</td>
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<tr>
<td>Vitamin B1 (as Thiamine Mononitrate)</td>
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<td>Potassium</td>
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<tr>
<td>Vitamin B2 (as Riboflavin)</td>
<td>1.7mg</td>
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<td>Vitamin B3 (as Niacinamide)</td>
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<td>Vitamin B5 (as D-Calcium Pantothenate)</td>
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<td>Vitamin B6 (as Pyridoxine HCL)</td>
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<td>Vitamin B12 (as Cyanocobalamin)</td>
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<tr>
<td>Folic Acid</td>
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<tr>
<td>Biotin</td>
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<tr>
<td>Calcium (as Calcium Carbonate)</td>
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<tr>
<td>Phosphorus</td>
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<td>Selenium</td>
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*Other Ingredients: Stearic Acid, Acacia Gum, Microcrystalline Cellulose, Magnesium Stearate, Silicon Dioxide.*
Percentage of Population that Live on Less than $1 a Day

A bottle of Centrum costs around $20

World Hunger Education Service
How Scientists use these Models to Understand Congenital Defects

1. What are you interested in testing?
2. Perturb the system and observe the experiment!
Vitamin A Inhibitors Block Normal Development (Congenital Defect)
1. Understand normal embryonic growth
2. Apply the scientific approach to discover the origin of congenital defects
3. Quantify the results using qualitative and quantitative observations