Down Syndrome
Genetic Counseling

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Down Syndrome (Trisomy 21)

- Incidence 1:650 births
- Strongly associated with increased maternal age, but can occur at any age
- Always genetic, but only a small proportion inherited (trisomy 21 (95%) vs translocation)
- Characteristic physical features
- Cardiac anomalies (50%)
- Developmental delays
- Risk for immunodeficiency, Alzheimer-like sx
The Genetics Evaluation

- History and Pedigree
- Physical Examination
- Laboratory Studies
- Counseling and Intervention
Epicanthal folds/ Brushfield spots
Large tongue/Flat nasal bridge
Single palmar crease
Sandal toe gap
Physical, Mental, and Social Characteristics of Down's Syndrome

Figure 22. Early development performance of Down's syndrome children raised at home compared to that of normal children. The widest point in each diamond represents the average age for performance, and the spread of the diamonds represents the range.
Genetic Testing

HUMAN CELL

46 Chromosomes (carry genes)

Nucleus
Down Syndrome
Etiologies

Trisomy 21    95%
Translocation 4%
Mosaic        1%
Translocation Down syndrome
## Parental Age

### Maternal

#### Down Syndrome Risk and Maternal Age

<table>
<thead>
<tr>
<th>Age (years)</th>
<th>Risk</th>
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<tbody>
<tr>
<td>&lt;25</td>
<td>1 / 1600</td>
</tr>
<tr>
<td>25 – 29</td>
<td>1 / 1100</td>
</tr>
<tr>
<td>30 – 34</td>
<td>1 / 700</td>
</tr>
<tr>
<td>35 – 39</td>
<td>1 / 250</td>
</tr>
<tr>
<td>40 – 42</td>
<td>1 / 80</td>
</tr>
<tr>
<td>&gt;42</td>
<td>1 / 40</td>
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Why has this happened?

Will it happen again?

What can be done about it?
Recurrence Risk

- Parents: 1 ½ % plus maternal age-related risk (unless there is a parental translocation, which makes the risk much higher)
- Other family members: same as the general population (unless translocation is running in the family)
- Individual with Down syndrome: generally infertile, but if they do have children, significant risk for a child with an extra chromosome
Prenatal diagnosis

- ACOG recommends that all women be offered aneuploidy screening during pregnancy, regardless of age.
- 1st trimester screen (ultrasound for nuchal translucency, blood for PAPP-A and B-hcg)
- Quad screen (AFP, hcg, estriol, DIA)
- Amniocentesis or chorionic villus sampling
- Ultrasound
Why do Prenatal Screening or Testing?

• Reasons are very personal and individual:
  – Reassurance
  – Improve obstetrical care
  – Cope with potential shock and grief prior to delivery
  – Plan for the future
  – Some women consider ending the pregnancy

• For certain women, this is unwanted information, and screening is not appropriate for them
Health care maintenance

- Echocardiogram at time of diagnosis
- Hearing and vision assessment
- Thyroid function tests
- Cervical spine X-ray
- Most other health supervision only as symptomatic, or similar to general pediatric population
- AAP “Health Supervision for Children with Down Syndrome”
Psychosocial Issues/Resources

• Down Syndrome Research Foundation
  www.dsrft.org

• National Down Syndrome Society
  www.ndss.org

• National Down Syndrome Congress
  www.ndscccenter.org

• Ollie Webb Center (ARC of Omaha, Career Solutions Inc.)
  www.olliewebbinc.org