Tetrasomy 18p

Reasoning is concrete-literal
Does better when concepts are simple and then expanded to include more complex ideas
No significant differences between age groups (Preschool, School age and Young adult)
Potential conditions in a neonate:
• Structural
  • Palatal anomalies - 80%
  • Heart abnormalities – 47% by ECG
  • Congenital orthopedic abnormalities – 45%
  • Hernias – 12%
  • Myelomeningocele – 7%
• Functional
  • Feeding problems – 83%
  • Respiratory distress – 31%
• Biochemical
  • Jaundice – 57%

Initial evaluations after diagnosis:
• Ophthalmology
  • Strabismus – 45%
  • Refractive errors – 34%
• Audiology / otolaryngology
  • Hearing loss - 32%
  • Recurrent otitis media – 35%
• Genitourinary
  • Cryptorchidism – 39%
  • Hypospadias – 4%
  • Urinary tract anomalies – 8%

Immediate Referrals to:
• Appropriate subspecialist as indicated by initial evaluations
• Genetics follow-up if not previous to diagnosis
• Early intervention/developmental services
• The Chromosome 18 Registry & Research Society
• The Chromosome 18 Clinical Research Center

Closely monitor and manage:
• Failure to thrive/ growth failure
  • Underweight (<3rd percentile)
• Endocrinology
  • Short stature (<25th percentile)
  • Growth hormone deficiency
• Otorhinolaryngology
  • Recurrent otitis media
  • Hearing loss
• Gastroenterology
  • Constipation
  • GE reflux
  • Hernias
  • Eosinophilic esophagitis
• Immunology/Rheumatology
  • Atopic disorders
  • IgA deficiency
  • Eosinophilic esophagitis
• Orthopedics
  • Congenital hip dysplasia
  • Foot abnormalities
  • Decreased bone mineral density
• Development
  • Milestones
  • School performance
• Neurology
  • Seizures
  • Hypotonia
• Behavioral/ mood changes
• Dental

Annual Screenings:
• Vision
• Hearing

Symptoms related to Eosinophilic Esophagitis (EoE)
• Feeding dysfunction. Coping mechanisms -avoiding highly textured foods such as meats and bulky foods such as bagels, cutting food in small pieces, lubricating foods before eating with liquids or butter, extensive chewing of foods, washing food down with liquids, prolongation of mealtimes
• Food impaction. Coping mechanisms - drinking liquid to wash food down, raising hands above head, jumping up and down, waiting for food to dissolve or to pass
• Chest pain. Coping mechanisms-avoiding foods or liquids that exacerbate pain such as highly textured or bulky foods, alcohol or acidic drinks
• GERD like symptoms recalcitrant to medical and surgical GERD management
• Abdominal pain
• Vomiting
• Anorexia and early satiety

Low bone density
• 54 responses (27 males, 27 females)
• 31% diagnosed with Low Bone Mineral Density, Osteopenia or Osteoporosis; 9 male, 8 female, 49 – 4 years old. Only 29% were being supplemented with calcium, vitamin D, or bisphosphonates.
• 59% had history of a broken bone; 41% Male, 44% Female (typical is 30-50%)
For more information see:
### Behavior: Tetrasomy 18p Adults

**Common problems**
- Performing basic tasks safely: 61%
- Expressing ideas and communication: 61%

**Less problems identified**
- Anti-social and rule-breaking behaviors: 11%
- Hostile actions: 6%
- Nervous, fearful, and worrisome tendencies: 6%
- Incapacitating sadness or stress: 6%
- Being overly sensitive about minor problems: 6%
- Inability to adapt readily to changes: 6%

### Social Impairment

**Common Problems**

<table>
<thead>
<tr>
<th>Adults</th>
<th>&gt;2.5 yrs.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Interpreting social cues</td>
<td>84%</td>
</tr>
<tr>
<td>Obsessing over same routines</td>
<td>84%</td>
</tr>
<tr>
<td>Picking up on social cues</td>
<td>68%</td>
</tr>
<tr>
<td>Social communication</td>
<td>68%</td>
</tr>
<tr>
<td>Motivation to engage in social behavior</td>
<td>47%</td>
</tr>
</tbody>
</table>

### Executive Functioning: Adults

**Common problems**
- Remembering information to complete a task: 89%
- Keeping track problem solving successes or failures: 83%
- Independently beginning a task: 72%
- Managing current and future oriented task demands: 67%
- Acting on an impulse: 61%

**Less common problem**
- Modulating emotional response: 6%

### Natural History

**Living Situation (>18 years)**
- Home with parents: 16
- With parents in independent area: 1
- Apartment (alone): 1
- Supervised independent living program: 1
- Group Home / Foster Home: 2
- Unknown: 3

**Highest Educational Level**
- Did Not Complete High School: 5
- Attending High School: 3
- High School Graduate (certificate): 9
- High School Graduate (diploma): 2
- Vocational School (certificate/degree): 1
- Some College/University (no degree): 1
- Unknown: 3

**Marital Status/Children**
- Married (no): 21
- Married (unknown): 3
- Children (none): 21
- Children (unknown): 3

**Work (Volunteer, Part-time, Full-time):**
- Yes: 12
- No: 9
- Unknown: 3

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**2016 Bleeding/Clotting study**

EMILIN2 - ?? Or any of 20 genes

*Our data: 11 18p- = poor platelet function
& 8 Tet18p = normal platelet function*
2 examples of bone density reports with the information we need

The reports we need should have the following:

- Images of the bone with lines indicating the region measured
- The Z-Score calculations for the appropriate age of the patient

We need these measurements for the:

- Femoral neck
- Total hip
- Lumbar spine
### Tetrasomy 18p: Current Ages & Age at Death

<table>
<thead>
<tr>
<th>Age deceased</th>
<th>Gender</th>
<th>Cause of death</th>
<th>Past medical history</th>
</tr>
</thead>
<tbody>
<tr>
<td>13 years 5 months</td>
<td>F</td>
<td>Sudden heart arrest. One day of history of nausea, vomiting and lethargy. Autopsy: significant dilatation of her colon</td>
<td>Significant constipation; question of seizures; a small VSD that closed on its own; broken bones (prone to accidents); left peroneal nerve palsy; osteoporotic ?; Achilles lengthening</td>
</tr>
<tr>
<td>3 years 5 months</td>
<td>F</td>
<td>n/a</td>
<td>Intrauterine growth retardation; static encephalopathy; severe gastroesophageal reflux; G-tube; horseshoe kidney; scoliosis; medical records available only as a an infant</td>
</tr>
</tbody>
</table>