An overview of CHARGE

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CHARGE Syndrome

• First identified in 1979
• It is a rare and challenging condition (both medically and educationally)
• Among genetic conditions it is now recognised as the leading cause of congenital deafblindness
• Is a low incidence disability affecting between 1:10,000 – 1:15,000 live births
## Major diagnostic characteristics of CHARGE syndrome

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Coloboma of the eye:</td>
<td>80-90%</td>
</tr>
<tr>
<td>Choanal atresia or stenosis</td>
<td>50-60%</td>
</tr>
<tr>
<td>Cranial nerve dysfunction or anomaly</td>
<td></td>
</tr>
<tr>
<td>• I – hyposmia or anosmia</td>
<td>&gt;90%</td>
</tr>
<tr>
<td>• VII – facial palsy (unilateral or bilateral)</td>
<td>40%</td>
</tr>
<tr>
<td>• VIII – hypoplasia of auditory nerve</td>
<td>&gt;80%</td>
</tr>
<tr>
<td>• IX/X – swallowing difficulties with aspiration</td>
<td>70-90%</td>
</tr>
<tr>
<td>Characteristic CHARGE outer ear</td>
<td>&gt;50%</td>
</tr>
<tr>
<td>Characteristic CHARGE middle or inner ear</td>
<td></td>
</tr>
<tr>
<td>• Ossicular malformations</td>
<td>&gt;80%</td>
</tr>
<tr>
<td>• Mondini defect of the cochlea</td>
<td>&gt;80%</td>
</tr>
<tr>
<td>• Absent/hypoplastic semicircular canals</td>
<td>&gt;90%</td>
</tr>
</tbody>
</table>

Ref: Hartshorne, Hefner, Davenport & Thelin (2011)
## Minor diagnostic characteristics of CHARGE syndrome

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genital hypoplasia</td>
<td>50%</td>
</tr>
<tr>
<td>Delayed puberty</td>
<td>&gt;50% in females/90% in males</td>
</tr>
<tr>
<td>Cardiovascular malformation</td>
<td>75-85%</td>
</tr>
<tr>
<td>Growth deficiency (short stature)</td>
<td>70%</td>
</tr>
<tr>
<td>Orofacial cleft (cleft lip and/or palate)</td>
<td>15-20%</td>
</tr>
<tr>
<td>Tracheoesophageal (T-E) fistula</td>
<td>15-20%</td>
</tr>
<tr>
<td>Renal anomalies</td>
<td>30-40%</td>
</tr>
<tr>
<td>Distinctive facial features</td>
<td>70-80%</td>
</tr>
<tr>
<td>Palmar crease</td>
<td>50%</td>
</tr>
<tr>
<td>CHARGE behavioural profile (including perseverative behaviours)</td>
<td>&gt;50%</td>
</tr>
</tbody>
</table>
CHD7 Gene
“CHARGE may be the only disorder that presents with deficits of all the senses.

Hartshorne et al, 2011
Some other aspects of CHARGE identified in literature

• Sensory integration
• Self-regulation
• Motor development
• Developmental milestones
• Adaptive behaviours
• Cognitive development
• Sleep disorders
• Altered perception of pain
• Growth and sex hormones
• Language development and communication
• Eating and drinking
• Behaviour
• Neurological impairments
• OCD/ ADHD/ ASD
• Issues developing with age
“I would like people to know I am not my disability”
CHARGE questionnaire

• Build upon our existing knowledge of CHARGE syndrome
• Help recognise areas of need
• Improve the support we can give to the children, young people and their families and to the professionals working with them.
Age of diagnosis

- 22/44 (55%) diagnosed with CHARGE before the age of 3 months
- 42/44 (95%) diagnosed before the age of 18 months
- Remaining 4 (9%) children were diagnosed before the age of 4 years
Diagnostic criteria
## Medication

<table>
<thead>
<tr>
<th>Number of medications</th>
<th>Number of children prescribed this number of medications</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>8</td>
</tr>
<tr>
<td>2</td>
<td>6</td>
</tr>
<tr>
<td>3</td>
<td>10</td>
</tr>
<tr>
<td>4</td>
<td>2</td>
</tr>
<tr>
<td>5</td>
<td>1</td>
</tr>
<tr>
<td>6</td>
<td>4</td>
</tr>
<tr>
<td>More than 6</td>
<td>2</td>
</tr>
</tbody>
</table>
Hospitalisation

29/44 (66%) babies in hospital for more than 2 weeks:

• 28/29- with feeding difficulties
• 22/29- for surgical intervention
• 27/29- for other medical reasons

• 17/29- because of a combination of feeding difficulties, surgical intervention and other medical reasons
Educational provision

Pre-School provision

- Early years class at state school
- Pre-school
- Opportunity group
- Specialist children’s centre
- Independent nursery
- State nursery for deaf children
Educational provision

School provision

- School for VI/ Blind
- School for children with physical difficulties
- School for the deaf
- School for children with Severe Learning Difficulties
- School for children with Severe Learning Difficulties / VI
- Non-maintained school
- Mainstream school
- Mainstream school with specialist VI or HI provision

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Statement of special educational needs