The incidence of childhood Gonadoblastoma over fifteen years in the Republic of Ireland

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BACKGROUND

• Gonadoblastoma (GB) is a rare tumour of the gonads presenting in childhood or adolescence.
• It is a lesion composed of a mixture of germ cells at different stages of maturation, with low malignant potential.
• It is associated with disorders of sex development (DSD), most commonly Turner mosaic syndrome with Y chromosome material (TMSY), and 46XY gonadal dysgenesis (GD).
• Little is known about the natural history and incidence of this rare tumour.

OBJECTIVES

• To determine the incidence and clinical features of GB presenting before age 16 years in the Republic of Ireland (RoI) over the fifteen year period from 1999 to 2013 inclusive.

METHODS

• A retrospective review of children and adolescents with a diagnosis of GB.
• Records of the National Cancer Registry Ireland and Departments of Endocrinology, Pathology and Surgery at the main paediatric units nationally.
• All children in RoI requiring gonadectomy are referred to a single tertiary referral centre thus anticipating good case ascertainment.
• Clinical notes and pathology reviewed.
• Incidence rate calculated using total number of cases (n=8) per total live births over the fifteen year period (n= 990,425) published by Central Statistics Office and expressed as rate per 10,000 live births.

RESULTS

Table 1: Patient characteristics including underlying diagnosis, age at gonadectomy and histopathology.

<table>
<thead>
<tr>
<th>Patient</th>
<th>Year of diagnosis of GB</th>
<th>Background diagnosis</th>
<th>Age at surgery</th>
<th>Histopathology</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>2000</td>
<td>TMSY</td>
<td>5 yrs</td>
<td>Ovotestis with early GB</td>
</tr>
<tr>
<td>2</td>
<td>2007</td>
<td>TMSY</td>
<td>14 yrs</td>
<td>Left sided GB. A small collection of cells on right ovary strongly suspicious of GB</td>
</tr>
<tr>
<td>3</td>
<td>2013</td>
<td>TMSY</td>
<td>6 m</td>
<td>Right gonad replaced by GB without evidence invasive germ cell tumours</td>
</tr>
<tr>
<td>4</td>
<td>2013</td>
<td>TMSY</td>
<td>13 m</td>
<td>Early GB</td>
</tr>
<tr>
<td>5</td>
<td>2013</td>
<td>TMSY</td>
<td>6 yrs</td>
<td>Extensive GB</td>
</tr>
<tr>
<td>6</td>
<td>2011</td>
<td>46XY GD</td>
<td>8 yrs</td>
<td>Right dysgerminoma (Fig 1a-b) Left GB (Fig 1c-e)</td>
</tr>
<tr>
<td>7</td>
<td>2012</td>
<td>46XY GD</td>
<td>4 m</td>
<td>Right testicular tissue and early GB Left fibrous gonad</td>
</tr>
<tr>
<td>8</td>
<td>2012</td>
<td>46XY GD</td>
<td>9 yrs</td>
<td>Early GB</td>
</tr>
</tbody>
</table>

• 8 cases of GB identified over the 15 year period (Table 1).
• All phenotypically female. All but one case GB was diagnosed on elective gonadectomy.
• 5 cases had Turner mosaic syndrome with Y chromosome material (TMSY), (age range 6 months – 14 years at diagnosis of GB), bilateral in 2 cases.
• Three cases of 46XY GD were aged 4 months, 8 and 9 years at diagnosis of GB.
• In only one case, a girl with 46XY GD with SRY deletion, clinical suspicion at age 8 prompted gonadectomy. Histology showed unilateral dysgerminoma and contralateral GB.

CONCLUSIONS

• The population incidence of childhood GB in the RoI is 0.08/10,000 births for the past 15 years.
• To our knowledge this is the first reported population incidence rate of childhood gonadoblastoma.
• Incidence may be underestimated due to other TMSY patients not being referred for gonadectomy.
• Due to the low age of GB in this cases series, the recommendation for elective gonadectomy in high risk conditions such as TMSY and 46XY GD is supported by this data.