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.

REFERENCES

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RESULTS

Table 1: Patient characteristics including underlying diagnosis, age at gonadectomy and histopathology.
TMSY = Turner mosaic syndrome with Y Chromosome material, GD = Gonadal Dysgenesis, GB = Gonadoblastoma

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

- 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.

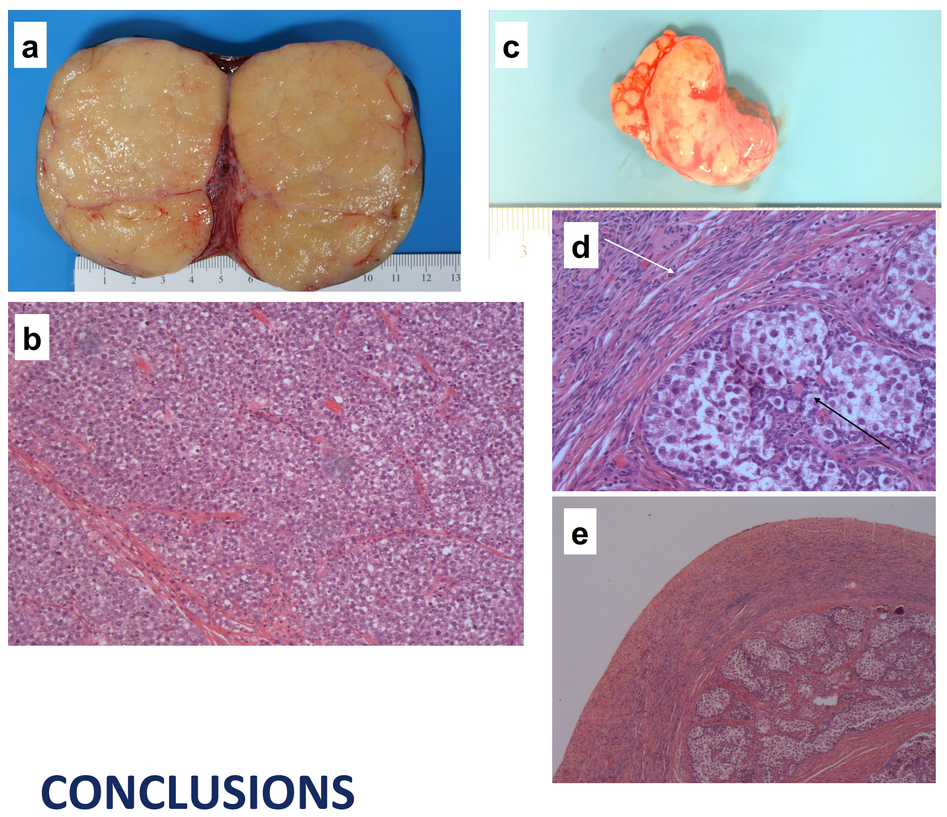

Figure 1: Patient 6 macropathology and histopathology specimens.
(a.) Right ovary opened up (b.) Right ovary: sheet-like growth of cells with pale eosinophilic cytoplasm, rounded vesicular nuclei and small nucleoli and showing high mitotic activity, features of germinoma
(c.) Left ovary (d.) Left ovary: streak gonad features on upper left (white arrow); residual gonadoblastoma (black arrow) lower right of slide.
(e.) Left streak gonad at periphery enclasping gonadoblastoma

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.