

Early Occurrence of Gonadoblastoma Found at Elective Gonadectomy in Turner Syndrome Mosaic for Y Chromosome

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BACKGROUND

Turner Syndrome (TS) is one of the most common genetic disorders in females and occurs in phenotypic females who are missing all or part of one sex chromosome.

Most common mosaic forms are 45,X/46,XX and 45,X/46,Xiq, and mosaicism for cells containing Y chromosome material is well documented.

Due to increased risk of gonadoblastoma (GB), current recommendations are for elective gonadectomy in girls with Turner mosaic syndrome with Y chromosome material (TMSY), following diagnosis.

However, recommendations on lower age limits for timing of elective gonadectomy in this situation are lacking.

METHODS

- Review of TS patients attending the Paediatric Endocrinology clinic in 2013 specifically looking at those with mosaicism for Y chromosome.

RESULTS

- Three of nine TS patients had TMSY (33% of TS clinical cohort).
- All had gonadoblastoma at time of elective gonadectomy at a young age.

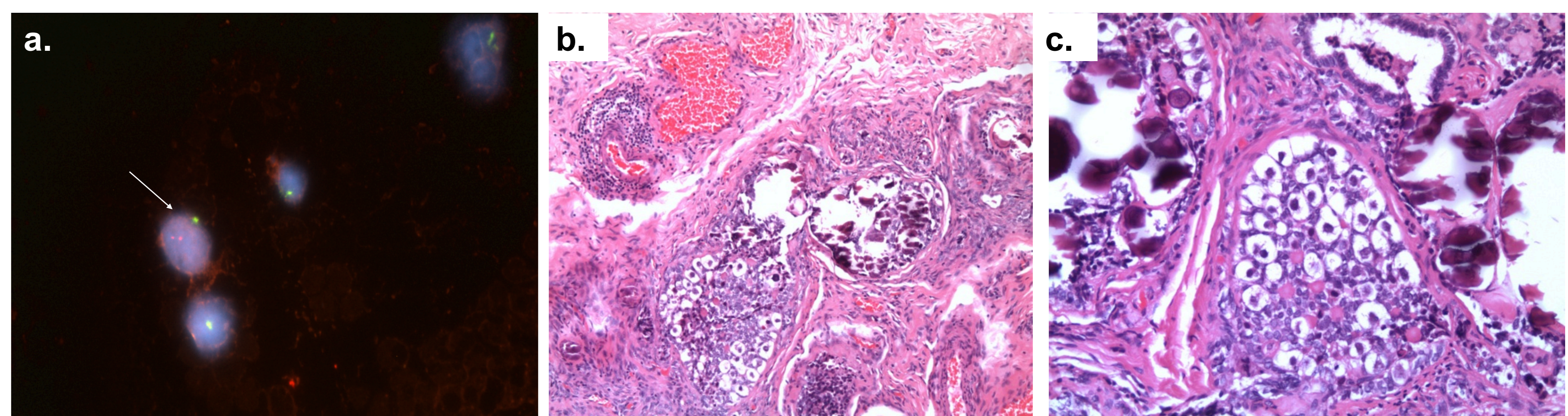
CONCLUSIONS

- This highlights the early age of occurrence of GB despite low mosaicism for SRY cell lines.
- Supports a recommendation for early surgery in such cases, regardless of age.

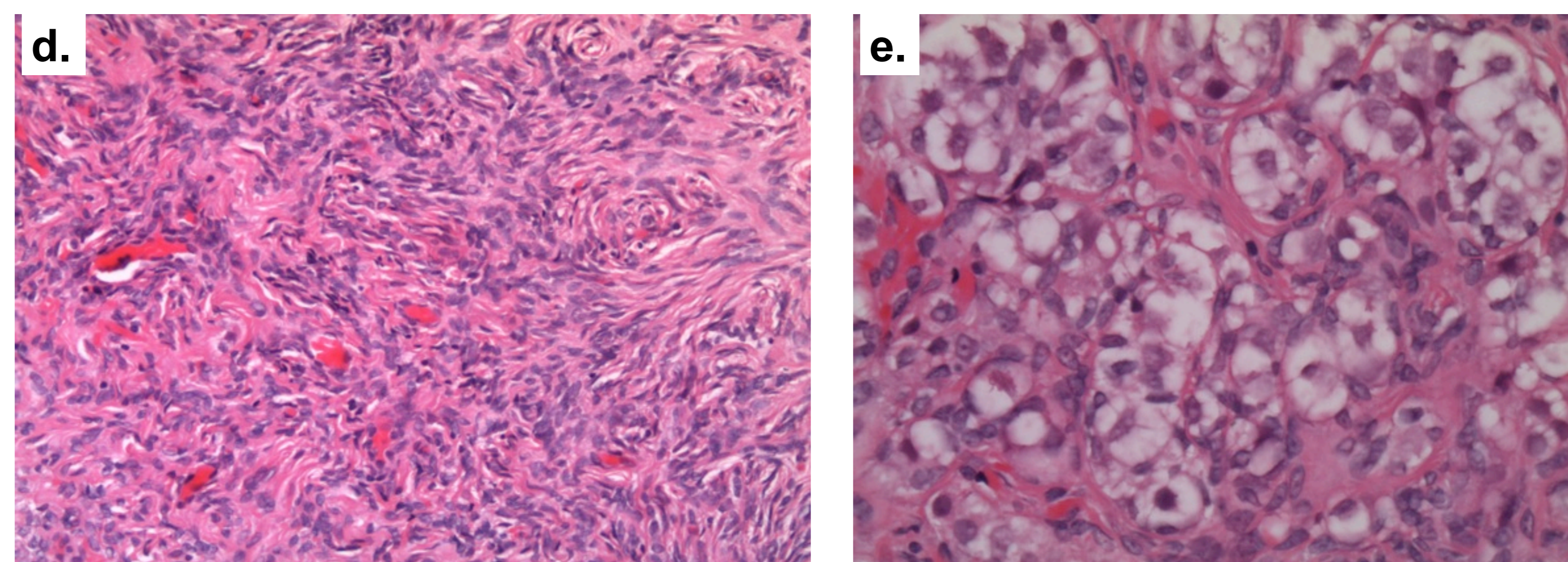
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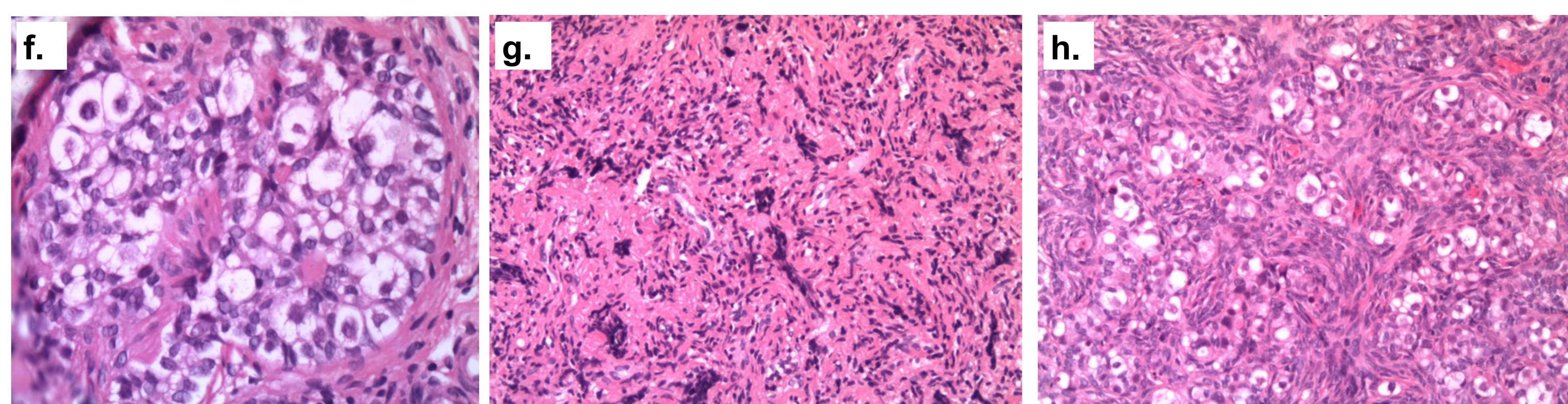
Case	Age at diagnosis TS	Presentation	Karyotype	Age surgery	Appearance at Resection	Histopathology
1	2 yrs	Routine paternity testing	45,X, 46,XY Blood 50 cell lines: 45,X (50%)/46,X, idic(Y)(q11.23).ish idic(Y)(q11.23)(SRY++) (50%) Buccal 45,X (100% cells) Gonad 45,X (57% cells) 46,X, idic(Y)(q11.23) (43% cells)	6 yrs	Bilateral streak gonads rudimentary mid-line uterus structure	Unilateral extensive GB
2	Birth	Dysmorphic features	45,X, 47,XY+18 Blood 200 cell lines: 45,X (95%)/47,XY+18 (5%) Buccal 45,X (100% cells) Gonad 45,X (86% cells)/47,XY,+18 (14% cells)	13 m	Bilateral streak ovaries Bilateral fallopian tubes with unilateral ectopic adrenal tissue	Bilateral early GB
3	Birth	Aortic stenosis	45,X, 46,XY Blood 35 cell lines: 45,X (83% cells)/46,X,psu idic(Y)(q11.23).ish(Y)(SRY++) (17% cells) Buccal 200 cell lines: nuc ish (DXZ1x1,SRYx0) (81%)/(DXZ1x1,SRYx2)(19%) Skin 230 cell lines: 45,X (13%)/46,X,psu idic(Y)(q11.23)[1 cell].nuc ish(SRYx0)(79%)/(SRYx2) (8%) Gonad karyotype not available	6 m	Bilateral gonads and fallopian tubes Small midline uterine structure	Unilateral extensive GB



Case 1: (a) Gonadal touch preparation fluorescence in situ hybridisation (FISH) study. Cell (white arrow) with green spot representing X chromosome, and 2 red spots representing SRY, reflecting an isochromosome Yp, The other two cells have 1 green spot, representing the 45,X cells (b) & (c) gonadoblastoma with characteristic almost organoid mixture of germ cell and sex cord elements.



Case 2: Histopathology (d) Streak gonad devoid of germ cells (e) gonadoblastoma



Case 3: Histopathology (f) microscopic gonadoblastoma (g) streak gonad (h) undifferentiated gonadal tissue with chaotic distribution of immature germ cells.