A Child with Clinical and Cytogenetic Features of Male Edward Syndrome and Turner Syndrome with Bilateral Gonadoblastoma in Infancy

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BACKGROUND

- Mosaic Turner Syndrome (TSM) commonly occurs in the form of 45,X/46,XX and 45,X/46,Xiq
- Mosaicism for the presence of a Y chromosome (45,X/46,XY) is well documented in TS.
- The presence of a Y chromosome in TS is associated with increased risk of gonadoblastoma (GB).
- To date, there are only 6 reported cases of TSM with a trisomy 18 karyotype, and only 2 of these were phenotypically female with 45,X, 47,XY+18 karyotype.

CASE REPORT

- Dysmorphic features noted at birth: webbed neck, low set ears and broad chest. The child had female external genitalia.
- Blood: G banded karyotype and interphase fluorescence in situ hybridisation (FISH) showed 45,X in 95% and 47,XY + 18 (Edwards Syndrome) in 5% of cells analysed.
- Buccal swab: Interphase FISH showed only 45,X (100% cells).
- Elective gonadectomy performed at 13 months of age.
- Bilateral streak ovaries with early evidence of GB bilaterally (Figure 2), rudimentary uterus and bilateral fallopian tubes with unilateral ectopic adrenal tissue.
- Gonadal tissue touch preparation: Interphase FISH similar to the blood with 45,X in 86% of cells and 47,XY+18 in 14% of cells analysed (Figure 1).

CONCLUSIONS

- This case highlights a rare karyotype of TSM and Edwards Syndrome in the same patient.
- Current investigations are ongoing as to the possible causes for this unusual finding.
- This case was also associated with a finding of early evidence bilaterally of gonadoblastoma at a very young age.
- Blood and gonadal karyotypes showed different levels of mosaicism.
- Mosaicism was limited, with the male Edward’s karyotype not detected in buccal cells.
- To our knowledge this is the only case with the above karyotype with early gonadoblastoma.

REFERENCES