Background

- The hCG stimulation test is a valuable method for assessing androgen production but there is a need to explore its utility in assessing androgen responsiveness and long-term prognosis

- Our aim was to explore the effect of hCG stimulation on the peripheral transcriptome in boys undergoing investigation for DSD

Methods

- 13 boys undergoing investigation for 46XY DSD received IM hCG 1500u on 3 consecutive days and had blood sampling on D0 and D3

- RNA was extracted from peripheral blood mononuclear cells on the Qiacube using RNA Blood Mini Kit with an incorporated DNase step. Microarray hybridisation was performed on 13 paired samples using the Affymetrix Human Transcript Array (HTA) 2.0

- Gene expression fold change was calculated and corrected for those boys who did not have a testosterone rise

Results

- Median age (range) at test was 0.83yrs (0.18-11.23) with a median External Masculinisation Score of 9 (6-11)

- 3 boys had isolated proximal hypospadias, 6 had bilateral undescended testes and 4 had a combination of hypospadias, impalpable testes or micropenis

- Median pre and post hCG testosterone were <0.5 nmol/l (<0.5-6) and 7.9 nmol/l (<0.5-31.5), respectively

- Median fold change of testosterone was 6.8 (1-26.6) and 3 (23%) boys did not demonstrate a testosterone rise (non-responders)

- Median AMH in the responders was 688 pmol/l (24, 1628) and in the non-responders was <4 pmol/l (<4, 256)

- White cell counts for the samples are shown in Figure 2

Conclusions

- The identification of a dynamic peripheral transcriptome that is associated with an androgen response following hCG stimulation extends the potential value of this clinical test

- The role of piRNAs as a diagnostic and prognostic marker of gonadal function needs further investigation

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