The prevalence of Turner Syndrome in girls who underwent surgical repair for Coarctation of Aorta; A 10 year single centre UK study

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Objective:
To identify whether genetic tests were performed in female infants who underwent surgical repair of Coarctation of aorta (CoA) and evaluate the occurrence of Turner syndrome.

Study design:
A total of 38 females who underwent surgical repair for CoA as a primary procedure in the last 10 years were identified. Patient data between March 2004 and January 2014 were analysed for genetic test results, availability of genetic counselling and prevalence of Turner syndrome in the patients.

Results
Of 38 females with CoA, 21 (55%) had karyotype analysis done. 3 out of the 21 patients were found positive for Turner syndrome. Genetic tests were not done for the rest 17 patients. This translated into a prevalence of 14.2% in the cohort with a minimum prevalence of 7.8% should the rest females be negative for Turner syndrome. All the patients who underwent genetic tests were offered counselling but 5 patients (28%) attended counselling.

Conclusion
Our study demonstrated for the first time in Europe that a minimum of 7.8% of girls presenting with CoA were found to have Turner syndrome on karyotyping. Turner syndrome has a huge spectrum of preventable and treatable health problems which can be addressed more effectively if diagnosed early. We propose that all girls with CoA should have a karyotype analysis at the time of diagnosis of CoA.