

ASHG#1136/T

Elisabeth de Jong, Hannie Douben, Bert Eussen, Dick Tibboel and Annelies de Klein
Department of Clinical Genetics and Pediatric Surgery, Erasmus MC, Rotterdam, the Netherlands

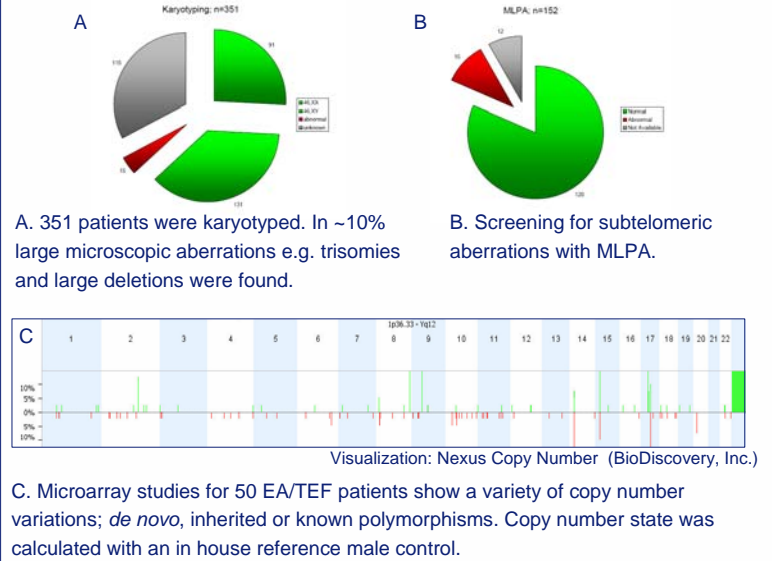
Introduction

The departments of Pediatric Surgery and Clinical Genetics have collected a large cohort of patients with tracheal and foregut-related anomalies. The most prevalent are Esophageal Atresia (EA) with or without Tracheo-Esophageal Fistula (TEF), associated in ~20% with VACTERL anomalies (Vertebral, Anal, Cardiovascular, Tracheo-Esophageal, Renal and Limb). Tracheal agenesis (TA), a congenital anomaly of the respiratory tract with an incidence of 1 in 50,000 births, is less frequent. We were able to collect 12 TA cases and DNA of 150 EA/TEF patients. Microarray studies were conducted to screen for small deletions or duplications.

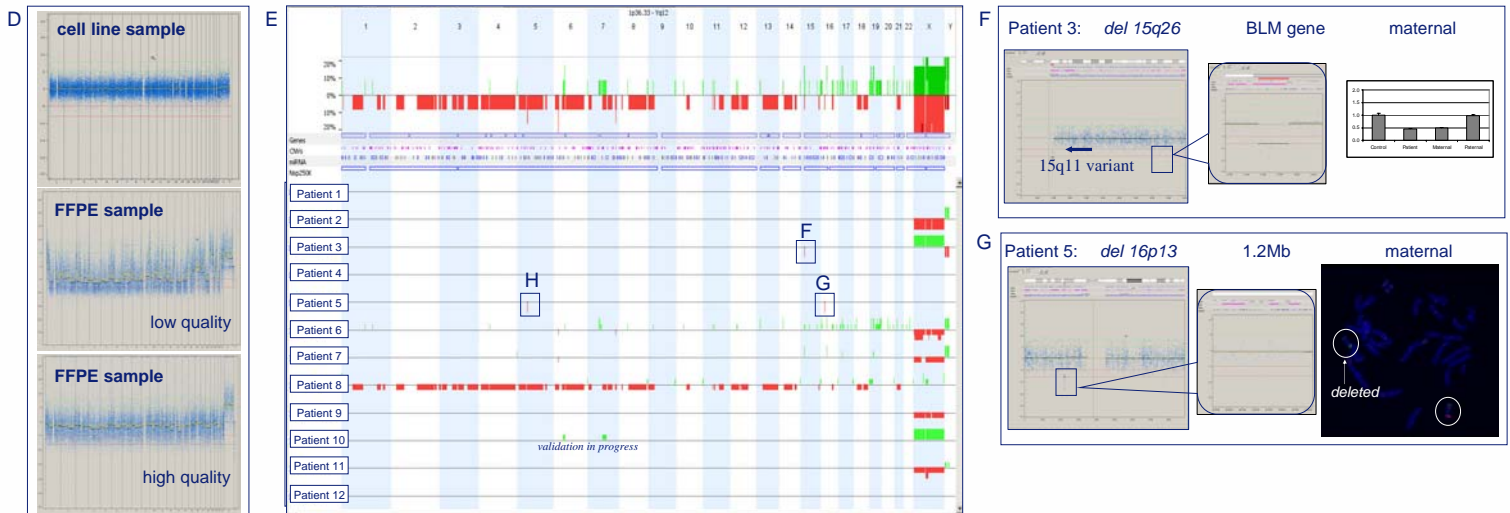
Material & Methods

Routinely, EA/TEF and TA patients are karyotyped and screened for subtelomeric aberrations with MLPA (Multiplex Ligation-dependent Probe Amplification). Array-CGH was used for paraffin embedded tissues (n=6) and cell lines (n=6) (105K Oligonucleotide-array; Agilent Technologies ®) and 50 EA/TEF patients were screened with Affymetrix® 250K NSP.

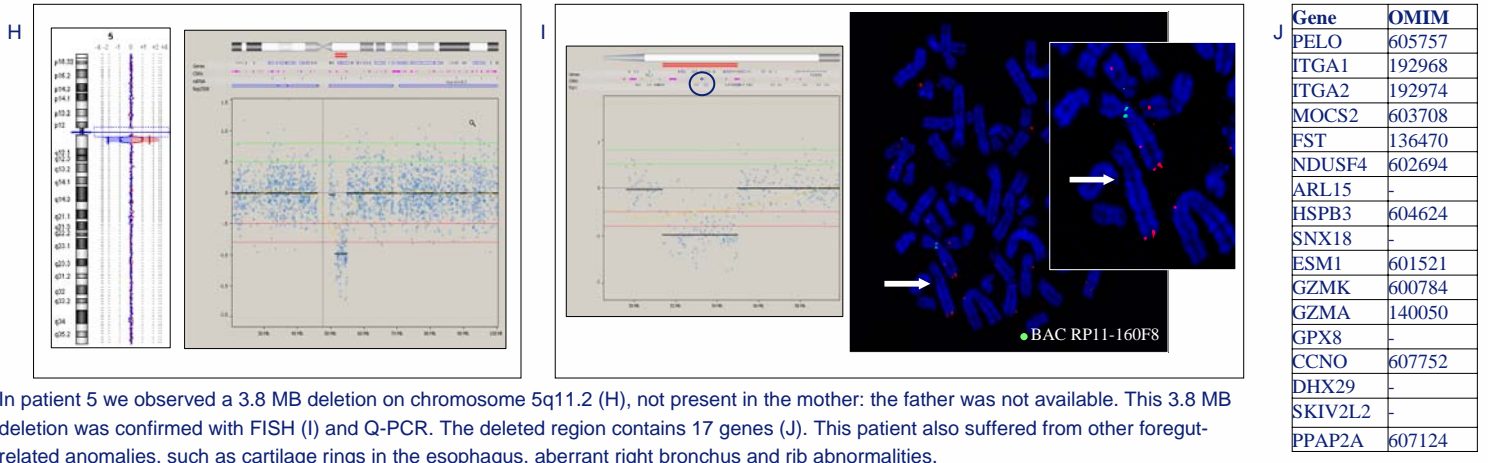
Results Molecular Cytogenetics



Results TA cohort



Results Patient 5



Conclusions

ArrayCGH is a comprehensive technique to study copy number variations in patients with congenital anomalies. Quality Array CGH data can be derived from FFPE samples with chemical labelling. In addition, new visualizing tools e.g. Nexus Copy Number (BioDiscovery, Inc.) provide additive analysing options of data generated on different platforms and compare those.