

Joint TECHGENE and EuroGentest (EUGT2) satellite meeting 'Innovative Techniques in Genome Diagnostics' ESHG 2012, Nürnberg, Germany

Monday June 25, 11.40-13.10, room St. Petersburg

Chair: Egbert **BAKKER**, LUMC, Leiden, NL (EuroGentest2) and
Hans **Scheffer**, Radboud UNMC, Nijmegen, NL (TECHGENE)

- 11:40- 11:55 Asuragen (AmplideX) technologies for FMR1 testing
Domenico **Coviello** and Marina **Grasso**,
Laboratory of Human Genetics, Genova, IT
- 11:55- 12:15 Quality aspects of diagnostic exome sequencing (TECHGENE)
Joep **de Ligt**, (Radboud UNMC, Nijmegen, NL)
- 12:15- 12:35 NGS quality aspects
Chris **Mattocks**, NGRL Salisbury, UK
- 12.35- 12.50 NGS data interpretation and validation challenges
Steven **Van Vooren**, Cartagenia, Leuven, BE
- 12:50 - 13:10 NGS/NIPD – Limits to the sensitivity of Non-Invasive Prenatal Diagnosis:
the importance of a minimal fraction of fetal DNA and the reduction of
overdispersion.
Gerard **te Meerman**, UMC Groningen, NL