

## Cytogenetics & Fluorescence *in situ* hybridization

### Cytogenetics

The cytogenetics laboratory at Geneunity provides a full range of constitutional and cancer cytogenetic services. Dr. Katy Phelan, the laboratory director, has more than 20 years experience. The laboratory is staffed by highly skilled cytogenetic technologists who are certified as clinical specialists in cytogenetics by the National Credentialing Agency.

### FISH

In addition to offering high quality chromosome analysis, the cytogenetics laboratory also specializes in fluorescence *in situ* hybridization (FISH). FISH, a molecular cytogenetic technique, enables the analysis of disease specific abnormalities as well as the chromosomal localization of specific DNA sequences. It is offered for the detection of cryptic rearrangements, microdeletion syndromes, aneuploidy and marker chromosome identification. In addition to the listed probes provided in house, our laboratory services are available for validation and testing of novel FISH probes.

### FISH probes

#### Constitutional

AneuVysion®: +13/+18+21 / X/Y  
 Angelman Syndrome  
 Cri-du-Chat Syndrome  
 DiGeorge/Velo-Cardio-Facial Syndrome  
 Kallmann Syndrome  
 Miller-Dieker Syndrome / Lissencephaly  
 Phelan-McDermid Syndrome, deletion 22q13  
 Prader-Willi Syndrome  
 Smith-Magenis Syndrome  
 SRY/CEP X  
 Steroid Sulfatase Deficiency  
 ToTelVysion: Subtelomeres  
 trisomy 21  
 Williams Syndrome  
 Wolf-Hirschhorn Syndrome

#### Oncology

ALK 2p23, anaplastic large cell lymphoma  
 AML1/ETO t(8;21)  
 API2/MALT1 t(11;18)  
 ATM deletion 11q22.3

BCL1 IGH/CCND1 t(11;14)  
 BCL2 IGH/BCL2 t(14;18)  
 BCL6 3q27 Rearrangement  
 BCR/ABL +9q34, t(9;22)  
 C MYC 8q24 Rearrangement  
 CBFβ t(16;16), inv(16)  
 CEP7 -7/7q31-  
 Deletion 13q14.3  
 Deletion 20q12  
 EGR1 -5/5q-  
 EGFR 7p12 Amplification  
 EWSR1 22q12 Rearrangement Ewing Sarcoma  
 FIP1L1/PDGFR4 4q12 Rearrangement  
 HER2/neu PathVysion®  
 IGH 14q32 Rearrangement  
 IGH/FGFR3 t(4;14)  
 IGH/MAF t(14;16)  
 IGH/MALT1 t(14;18)  
 IGH/MYC, CEP 8 t(8;14)  
 MALT1 18q21  
 MLL 11q23  
 MYB deletion 6  
 N-MYC 2p24.1  
 Oligodendroglioma 1p36-/19q13- Glioma  
 p53 deletion 17p13  
 PML/RARA t(15;17)  
 PTEN/CEP10 deletion 10q23  
 SYT 18q11.2 Synovial Sarcoma  
 TEL/AML1 t(12;21)  
 trisomy 3  
 trisomy 4  
 trisomy 5, 9 and 15  
 trisomy 8  
 trisomy 10  
 trisomy 12  
 UroVysion® (R) +3/+7/+17/deletion 9p21