

## Gestione ed Utilizzo di Biobanche Genetiche

### Dottorandi coinvolti XXII ciclo

#### 1° anno

- ❖ Baharak Khadang (*Genetica Medica*)  
“X-linked Alport syndrome: molecular and clinical aspects”
- ❖ Elena Marcocci (*Genetica Medica*)  
“Analysis of COL4A4 gene in autosomal Alport syndrome”
- ❖ Filomena Tiziana Papa (*Genetica Medica*)  
“Study of the WDR36 gene in patients with primary open angle glaucoma”
- ❖ Ariele Rosseto (*Genetica Medica*)  
“Analysis of PKD1 and PKD2 genes in Polycystic Kidney Disease”
- ❖ Rita Filippi (*Allergologia e immunologia cliniche e sperimentali*)  
“Assessment of a database for the follow-up of patients with pulmonary fibrosis”
- ❖ Camilla D’Eramo (*Meccanismi di neurodegenerazione, neuroprotezione e neuroriparazione delle malattie neurologiche rare*)  
“Mutations in NOTCH 3 and CADASIL: genotype-phenotype correlation”
- ❖ Cinzia Batisti (*Patologia degenerativa, involutiva e neoplastica oftalmologia*)  
“Corneal diseases”
- ❖ Giuliana Malagnino (*Patologie epatobiliopancreatiche e sindromi multitumorali*)  
“Extracolonic manifestations of Familial Adenomatous Polyposis”
- ❖ Daniela Giachino (*Genetica Medica, Università di Torino*)  
“Study of modifier factors in prostate and lung cancer”

#### 2° anno

- ❖ Rosangela Artuso (*Genetica Medica*)  
“Study of functional polymorphisms as modulating factors of Rett syndrome”
- ❖ Eleni Katzaki (*Genetica Medica*)  
“Clinical and molecular study of patients affected by Cohen syndrome”
- ❖ Federica Ottimo (*Genetica Medica*)  
“Analysis of the spectrum of MECP2 mutations in Rett syndrome”
- ❖ Tiziana Squillaro (*Genetica Medica*)  
“Role of the LRRK2 gene in Parkinson disease”
- ❖ Marina Vignoli (*Genetica Medica*)  
“Functional variants in genes involved in the metabolism of the 5-fluorouracile”
- ❖ Gianluca Martone (*Patologia degenerativa, involutiva e neoplastica oftalmologia*)  
*Keratoconus associated with corneal macular dystrophy*
- ❖ Patrizia Pitierri (*Patologia degenerativa, involutiva e neoplastica oftalmologia*)  
*Clinical approaches for orbital tumours*
- ❖ Lucia Russo (*Patologia degenerativa, involutiva e neoplastica oftalmologia*)  
*Clinical assessment in glaucoma patients*
- ❖ Francesco Scarpa (*Patologia degenerativa, involutiva e neoplastica oftalmologia*)  
*Clinical corneal confocal microscopi*
- ❖ Salvatore Spina (*Meccanismi di neurodegenerazione, neuroprotezione e neuroriparazione delle malattie neurologiche rare*)  
*Tau protein and dementia*
- ❖ Luca Cantarini (*Reumatologia sperimentale*)  
*Role of cytokines in autoinflammatory diseases*

- ❖ Angela Mazzi (*Allergologia e immunologia cliniche e sperimentali*)

Clinical assessment of patients with interstitial lung diseases

- ❖ Leonardo Barellini (*Patologie epatobiliopancreatiche e sindromi multitumorali*)  
Papillary carcinoma of the thyroid and FAP

### 3° anno

- ❖ Katia Sampieri (*Genetica Medica*)

Molecular determinants of variable expressivity in Retinoblastoma

- ❖ Vincenza Causarano (*Genetica Medica*)

Molecular study of premature ovarian failure

- ❖ Simona Albano (*Allergologia e immunologia cliniche e sperimentali*)

Assessment of a database for the follow-up of patients with sarcoidosis

- ❖ Maria Rosaria Martino (*Allergologia e immunologia cliniche e sperimentali*)

Study of pulmonary fibrosis associated with systemic sclerosis

- ❖ Sonia De Francesco (*Patologia degenerativa, involutiva e neoplastica oftalmologia*)

Late onset Retinoblastoma and prognosis

- ❖ Francesca Lepri (*Patologia degenerativa, involutiva e neoplastica oftalmologia*)

Genetic origins of cataract

- ❖ Marco Alegente (*Patologia degenerativa, involutiva e neoplastica oftalmologia*)

Methods for staining the anterior capsule

- ❖ Eugenia Borgione (*Meccanismi di neurodegenerazione, neuroprotezione e neuroriparazione delle malattie neurologiche rare*)

Mitochondrial DNA Mutations in syndromes with mental retardation

- ❖ Armand Dhamo (*Patologie epatobiliopancreatiche e sindromi multitumorali*)

Hilar versus common duct cholangiocarcinoma

- ❖ Federico Mariani (*Patologie epatobiliopancreatiche e sindromi multitumorali*)

Intraductal papillary mucinous tumours of Pancreas

### 4° anno

- ❖ Rossella Caselli (*Genetica Medica*)

Application of the array CGH technique on mentally retarded patients

- ❖ Elisa Scala (*Genetica Medica*)

Quantitative analysis of the MECP2 gene in Rett syndrome

- ❖ Gian Nicola Gallus (*Meccanismi di neurodegenerazione, neuroprotezione e neuroriparazione delle malattie neurologiche rare*)

Genotype-phenotype correlations in patients with cerebrotendinous xanthomatosis

- ❖ Andrea Bernini (*Patologie epatobiliopancreatiche e sindromi multitumorali*)

Study of anorectal melanoma

- ❖ Paolo Motolese (*Patologia degenerativa, involutiva e neoplastica oftalmologia*)

Follow-up of patients with glaucoma

- ❖ Anna Maria Catanese (*Patologia degenerativa, involutiva e neoplastica oftalmologia*)

Clinical assessment of patients with strabismus