ALLEGATO N. 2/A

TITOLI E PUBBLICAZIONI VALUTABILI

PROCEDURA SELETTIVA PER IL RECLUTAMENTO DI N. 1 RICERCATORE A TEMPO DETERMINATO DI TIPOLOGIA A PER IL SETTORE CONCORSUALE 06/A1 -GENETICA MEDICA- SETTORE SCIENTIFICO-DISCIPLINARE MED/03 -GENETICA MEDICA- PRESSO IL DIPARTIMENTO DI MEDICINA MOLECOLARE DELL'UNIVERSITÀ DEGLI STUDI DI ROMA "LA SAPIENZA" BANDITA CON D.D. Prot. N. 108/2020 DEL 21.01.2020

La Commissione prende atto dei titoli per i quali sia stata presentata idonea documentazione ai sensi dell'art. 3 del bando.

CANDIDATO: BOTTILLO IRENE

VERIFICA TITOLI VALUTABILI:

1. Titolo: "Dottorato di ricerca in Genetica Medica" conseguito presso la Università di Roma la Sapienza. VALUTABILE
2. Titolo: "Specializzazione in Genetica Medica" conseguito presso la Università di Roma la Sapienza. VALUTABILE

VERIFICA PUBBLICAZIONI VALUTABILI

Titoli delle pubblicazioni

1. Identification of a variant hotspot in MYBPC3 and of a novel CSRP3 autosomal recessive alteration in a cohort of Polish patients with hypertrophic cardiomyopathy. VALUTABILE
2. Eleven novel SLC12A1 variants and an exonic mutation cause exon skipping in Bartter syndrome type I. VALUTABILE
3. Familial hypomagnesaemia, Hypercalciuria and Nephrocalcinosis associated with a novel mutation of the highly conserved leucine residue 116 of Claudin 16 in a Chinese patient with a delayed diagnosis: a case report. VALUTABILE
4. Biallelic variants in the ciliary gene TMEM67 cause RHYNS syndrome VALUTABILE
5. The novel mutation in exon 9 of Cullin 3 gene contributes to aberrant splicing in pseudohypoaldosteronism type II. VALUTABILE
6. A novel germline mutation in CDK4 codon 24 associated to familial melanoma. VALUTABILE
7. Functional Characterization of a Novel Truncating Mutation in Lamin A/C Gene in a Family with a Severe Cardiomyopathy with Conduction Defects. VALUTABILE
8. A recurrent deletion in the SLC5A2 gene including the intron 7 branch site responsible for familial renal glucosuria. VALUTABILE
9. A novel LAMP2 mutation associated with severe cardiac hypertrophy and microvascular remodeling in a female with Danon disease: a case report and literature review. VALUTABILE
10. Clinical and molecular characterization of a boy with intellectual disability, facial dysmorphism, minor digital anomalies and a complex IL1RAPL1 intragenic rearrangement. VALUTABILE
11. Prediction and visualization data for the interpretation of sarcomeric and non-sarcomeric DNA variants found in patients with hypertrophic cardiomyopathy. VALUTABILE
12. Molecular analysis of sarcomeric and non-sarcomeric genes in patients with hypertrophic cardiomyopathy. VALUTABILE
13. Two Novel HOGA1 Splicing Mutations Identified in a Chinese Patient with Primary Hyperoxaluria Type 3. VALUTABILE

14. p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. VALUTABILE
15. An additional patient with 3q27.3 microdeletion syndrome VALUTABILE
16. Disorders of sex development: a genetic study of patients in a multidisciplinary clinic. VALUTABILE
17. A 22-Week-Old Fetus with Nager Syndrome and Congenital Diaphragmatic Hernia due to a Novel SF3B4 Mutation. VALUTABILE
18. Prenatal diagnosis and post-mortem examination in a fetus with thrombocytopenia-absent radius (TAR) syndrome due to compound heterozygosity for a 1q21.1 microdeletion and a RBM8A hypomorphic allele: a case report. VALUTABILE
19. Familial spinal neurofibromatosis due to a multiexonic NF1 gene deletion. VALUTABILE
20. Mutations in PVRL4, encoding cell adhesion molecule nectin-4, cause ectodermal dysplasia-syndactyly syndrome. VALUTABILE
21. Germline mosaicism in neurofibromatosis type 1 due to a paternally derived multi-exon deletion. VALUTABILE
22. High-resolution SNP arrays in mental retardation diagnostics: how much do we gain? VALUTABILE
23. HDR (Hypoparathyroidism, Deafness, Renal dysplasia) syndrome associated to GATA3 gene duplication. VALUTABILE
24. Germline and somatic NF1 mutations in sporadic and NF1-associated malignant peripheral nerve sheath tumours. VALUTABILE
25. Array-based comparative genomic hybridization in early-stage mycosis fungoides: recurrent deletion of tumor suppressor genes BCL7A, SMAC/DIABLO, and RHOF. VALUTABILE
26. RAS signaling in colorectal carcinomas through alteration of RAS, RAF, NF1, and/or RASSF1A. VALUTABILE
27. Deletions of NF1 gene and exons detected by multiplex ligation-dependent probe amplification. VALUTABILE
28. Functional analysis of splicing mutations in exon 7 of NF1 gene VALUTABILE
29. NF1 gene mutations represent the major molecular event underlying neurofibromatosis-Noonan syndrome. VALUTABILE
30. Novel and recurrent mutations in the NF1 gene in Italian patients with neurofibromatosis type 1. VALUTABILE

TESI DI DOTTORATO
"Innovazioni Tecnologiche per lo studio della complessità del genoma umano" VALUTABILE

CONSISTENZA COMPLESSIVA DELLA PRODUZIONE SCIENTIFICA:
Il candidato presenta una produzione complessiva pari a n.42 pubblicazioni.

CANDIDATO: GUIDA VALENTINA

VERIFICA TITOLI VALUTABILI:

1. Titolo "Diploma di Specializzazione in Genetica Medica conseguito presso la Università di Roma la Sapienza". VALUTABILE.
2. Titolo "Dottore di Ricerca in Genetica Medica conseguito presso la Università di Roma la Sapienza". VALUTABILE.

VERIFICA PUBBLICAZIONI VALUTABILI
Titoli delle pubblicazioni

1. Reliability of DHPLC in Mutational Screening of b-Globin (HBB) Alleles". VALUTABILE

2. "Extrachromosomal genes: a powerful tool in gene targeting approaches". VALUTABILE

3. "Use of DHPLC for Rapid Screening of Recombinant Clones". VALUTABILE

4. "Molecular detection of novel WFS1 mutations in patients with Wolfram syndrome by a DHPLC-based assay". VALUTABILE

5. "Phylogeographic Analysis of Haplogroup E3b (E-M215) Y Chromosomes Reveals Multiple Migratory Events Within and Out Of Africa". VALUTABILE

6. "Denaturing HPLC-based assay for molecular screening of nondeletional mutations causing alpha-thalassemias". VALUTABILE

7. "Screening of mutations in the CFTR gene in 1195 couples entering assisted reproduction technique programs". VALUTABILE

8. "Hematologic and molecular characterization of a Sicilian cohort of a thalassemia carriers". VALUTABILE

9. "Influence of Gg –158 C®T and b- (AT)x(T)y globin gene polymorphisms on HbF levels in Italian b-thalassemia carriers and wild-type subjects". VALUTABILE

10. "Sequence-specific modification of a β-thalassemia locus by small DNA fragments in human erythroid progenitor cells". VALUTABILE

11. "A Functional Variant of the Adipocyte Glycerol Channel Aquaporin 7 Gene Is Associated With Obesity and Related Metabolic Abnormalities". VALUTABILE

12. "Interaction of DIO2 T92A and PPARG2 P12A Polymorphisms in the Modulation of Metabolic Syndrome". VALUTABILE

13. "Familial transposition of the great arteries caused by multiple mutations in laterality genes". VALUTABILE

14. "Multiplex ligation-dependent probe amplification analysis of GATA4 gene copy number variations in patients with isolated congenital heart disease". VALUTABILE

15. "Design of novel three-phase PCL/TZ–HA biomaterials for use in bone regeneration applications". VALUTABILE

16. "Application of MLPA assay to characterize unsolved α-globin gene rearrangements". VALUTABILE

17. "Ebstein Anomaly: Genetic Heterogeneity and Association With Microdeletions 1p36 and 8p23.1". VALUTABILE

18. "Novel and recurrent JAG1 mutations in patients with tetralogy of Fallot ". VALUTABILE

19. “A variant in the carboxyl-terminus of connexin 40 alters GAP junctions and increases risk for tetralogy of Fallot”. VALUTABILE

20. “JAG1 Mutation in a Patient With Deletion 22q11.2 Syndrome and Tetralogy of Fallot ”. VALUTABILE

21. “ Novel and recurrent EVC and EVC2 mutations in Ellis-vanCreveld Syndrome and Weyers acrofacial dyostosis”. VALUTABILE

22. “A De Novo Proximal 3q29 Chromosome Microduplication in a Patient with Oculo Auriculo Vertebral Spectrum”. VALUTABILE

23. “Oculodentodigital Dysplasia with Massive Brain Calcification and a New Mutation of GJA1 Gene”. VALUTABILE

24. “Identification of a second HOXA2 nonsense mutation in a family with autosomal dominant non-syndromic microtia and distinctive ear morphology”. VALUTABILE

25. “GLI1 inactivation is associated with developmental phenotypes overlapping with Ellis–van Creveld syndrome”. VALUTABILE

26. “Biallelic mutations in DYNC2LI1 are a rare cause of Ellis-van Creveld syndrome ”. VALUTABILE

27. Novel α-Actin Gene Mutation p.(Ala21Val) Causing Familial Hypertrophic Cardiomyopathy, Myocardial Noncompaction, and Transmural Crypts. Clinical-Pathologic Correlation. VALUTABILE

28. “Heterozygous missense mutations in NFATC1 are associated with atrioventricular septal defect”. VALUTABILE

29. “Delineation of MidXq28-duplication syndrome distal to MECP2 and proximal to RAB39B genes”. VALUTABILE

30. “A new case of SMABF2 diagnosed in stillbirth expands the prenatal presentation and mutational spectrum of ASCC1”. VALUTABILE

<u>TESI DI DOTTORATO:</u>

Titolo: “Approcci di terapia genica mediante correzione sito-specifica: la B-talassemia come modello di studio." VALUTABILE

CONSISTENZA COMPLESSIVA DELLA PRODUZIONE SCIENTIFICA:

Il candidato presenta una produzione complessiva pari a n. 41 pubblicazioni .

CANDIDATO: MANCINI CECILIA

VERIFICA TITOLI VALUTABILI:

1.Titolo: “Dottorato in Genetica Umana” conseguito presso l’Università di Torino. VALUTABILE

VERIFICA PUBBLICAZIONI VALUTABILI
Titoli delle pubblicazioni

1. Missense mutations in the AFG3L2 proteolytic domain account for ~1.5% of European autosomal dominant cerebellar ataxias. VALUTABILE
2. NT5E mutations and arterial calcifications. VALUTABILE
3. Gene-targeted embryonic stem cells: real-time PCR assay for estimation of the number of neomycin selection cassettes. VALUTABILE
4. Megalencephalic leukoencephalopathy with subcortical cysts type 1 (MLC1) due to a homozygous deep intronic splicing mutation. VALUTABILE
5. High frequency of ribosomal protein gene deletions in Italian Diamond-Blackfan anemia patients detected by multiplex ligation-dependent probe amplification assay. VALUTABILE
6. Spastic paraplegia gene 7 in patients with spasticity and/or optic neuropathy. VALUTABILE
7. Genome-wide expression profiling and functional characterization of SCA28 lymphoblastoid cell lines reveal impairment in cell growth and activation of apoptotic pathways. VALUTABILE
8. ELOVL5 mutations cause spinocerebellar ataxia 38. VALUTABILE
9. Adult-onset autosomal recessive ataxia associated with neuronal ceroid lipofuscinosis type 5 gene (CLN5) mutations. VALUTABILE
10. Two families with novel missense mutations in COL4A1: When diagnosis can be missed. VALUTABILE
11. Blood metal levels and related antioxidant enzyme activities in patients with ataxia telangiectasia. VALUTABILE
12. An atypical form of AOA2 with myoclonus associated with mutations in SETX and AFG3L2. VALUTABILE
13. A novel 3q29 deletion associated with autism, intellectual disability, psychiatric disorders, and obesity. VALUTABILE
14. Ribosomal RNA analysis in the diagnosis of Diamond-Blackfan Anaemia. VALUTABILE
15. Updated genetic testing of Italian patients referred with a clinical diagnosis of primary hyperoxaluria. VALUTABILE
16. Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proof-of-concept examples. VALUTABILE
17. RPL5 on 1p22.1 is recurrently deleted in multiple myeloma and its expression is linked to bortezomib response. VALUTABILE
18. Exome sequencing in children of women with skewed X-inactivation identifies atypical cases and complex phenotypes. VALUTABILE
19. A case of Feingold type 2 syndrome associated with keratoconus refines keratoconus type 7 locus on chromosome 13q. VALUTABILE
20. CNVs analysis in a cohort of isolated and syndromic developmental DD/ID reveals novel genomic disorders, position effects and candidate disease genes. VALUTABILE
21. A novel homozygous change of CLCN2 (p.His590Pro) is associated with a subclinical form of leukoencephalopathy with ataxia (LKPAT). VALUTABILE

22. Human canonical CD157/Bst1 is an alternatively spliced isoform masking a previously unidentified primate-specific exon included in a novel transcript. VALUTABILE
23. Spinocerebellar Ataxia Tethering PCR: A Rapid Genetic Test for the Diagnosis of Spinocerebellar Ataxia Types 1, 2, 3, 6, and 7 by PCR and Capillary Electrophoresis. VALUTABILE
24. Altered homeostasis of trace elements in the blood of SCA2 patients. VALUTABILE
25. Prevalence and phenotype of the c.1529C>T SPG7 variant in adult-onset cerebellar ataxia in Italy. VALUTABILE
26. ATXN2 intermediate repeat expansions influence the clinical phenotype in frontotemporal dementia. VALUTABILE
27. Mice harbouring a SCA28 patient mutation in AFG3L2 develop late-onset ataxia associated with enhanced mitochondrial proteotoxicity. VALUTABILE
28. A fetal case of microphthalmia and limb anomalies with abnormal neuronal migration associated with SMOC1 biallelic variants. VALUTABILE
29. Spontaneous remission in a Diamond-Blackfan anaemia patient due to a revertant uniparental disomy ablating a de novo RPS19 mutation. VALUTABILE

TESI DI DOTTORATO

Titolo: “Functional characterization of AFG3L2 missense mutations in Spinocerebellar Ataxia type 28 and development of a mouse model of the disease”. VALUTABILE

CONSISTENZA COMPLESSIVA DELLA PRODUZIONE SCIENTIFICA:

Il candidato presenta una produzione complessiva pari a n. 33 pubblicazioni (delle 34 dichiarate la pubblicazione n.19 è una immagine di copertina relativo alla pubblicazione n.18),

CANDIDATO: PEDACE LUCIA

VERIFICA TITOLI VALUTABILI:

1. Titolo: “Specializzazione in Genetica Medica” conseguito presso la Università di Roma la Sapienza. VALUTABILE.
2. Titolo: Dottorato in Genetica Medica VALUTABILE conseguito presso la Università di Roma la Sapienza. VALUTABILE

VERIFICA PUBBLICAZIONI VALUTABILI
Titoli delle pubblicazioni:

1. Modeling Medulloblastoma in Vivo and With Human Cerebellar Organoids. VALUTABILE
2. Frameshift Mutations at the C-terminus of HIST1H1E Result in a Specific DNA Hypomethylation Signature. VALUTABILE
3. Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. VALUTABILE
4. Role of DNA Methylation Profile in Diagnosing Astroblastoma: A Case Report and Literature Review
   VALUTABILE
5. A novel germline mutation in CDK4 codon 24 associated to familial melanoma VALUTABILE
6. MFN2 transcripts escaping from nonsense-mediated mRNA decay pathway cause Charcot-Marie-Tooth disease type 2A2. VALUTABILE

7. Late-Onset Spastic Paraplegia Type 10 (SPG10) Family Presenting with Bulbar Symptoms and Fasciculations Mimicking Amyotrophic Lateral Sclerosis VALUTABILE
8. ALS5/SPG11/KIAA1840 mutations cause autosomal recessive axonal Charcot- Marie-Tooth disease. VALUTABILE
9. Longitudinal hormonal evaluation in a patient with disorder of sexual development, 46,XY karyotype and one NR5A1 mutation. VALUTABILE
10. A novel variant in the 3' untranslated region of the CDK4 gene: interference with microRNA target sites and role in increased risk of cutaneous melanoma. VALUTABILE
11. Comedonal Darier disease: report of 2 cases. VALUTABILE
12. Analysis of the miR-34a Locus in 62 Patients with Familial Cutaneous Melanoma Negative for CDKN2A/CDK4 Screening. VALUTABILE
13. Clinical Features Predicting Identification of CDKN2A Mutations in Italian Patients with Familial Cutaneous Melanoma. VALUTABILE
14. AXIN2 germline mutations are rare in 5 familial melanoma. VALUTABILE
15. Molecular Characterization of 11 Italian Patients with Darier Disease VALUTABILE
16. Jejunal atresia and anterior chamber anomalies: Further delineation of the Strømme syndrome. VALUTABILE
17. Fontaine-Farriaux syndrome: a recognizable craniosynostosis syndrome with nail, skeletal, abdominal, and central nervous system anomalies. VALUTABILE
18. A novel heterozygous SOX2 mutation causing anophthalmia/microphthalmia with genital anomalies. VALUTABILE
19. Increasing the BCR-ABL expression levels and/or the occurrence of ABL point mutations does not always predict resistance to Imatinib Mesylate in BCR-ABL positive acute lymphoblastic leukemia. VALUTABILE
20. Identification of a novel duplication in the APC gene using multiple ligation probe amplification in a patient with classic FAP. VALUTABILE

TESI DI DOTTORATO

Non dichiarato il titolo NON VALUTABILE

CONSISTENZA COMPLESSIVA DELLA PRODUZIONE SCIENTIFICA:

Il candidato presenta una produzione complessiva pari a n. 21 pubblicazioni,

La Commissione termina i lavori di verifica dei titoli dei candidati alle ore 18.30

Letto, approvato e sottoscritto.

LA COMMISSIONE

- Prof. Liborio Stuppia

- Prof. Emiliano Giardina

- Prof. Monica Rosa Miozzo