



DIPARTIMENTO DI SCIENZE MEDICHE E CHIRURGICHE

Modulo richiesta assegno

TUTOR	Annalisa Astolfi		
PRODUZIONE SCIENTIFICA TUTOR			
Punteggio VRA	1		

Commissione proposta 3 commissari + 1 supplente	Astolfi Annalisa
	Pantaleo Maria Abbondanza
	Nannini Margherita
	Lamberti Giuseppe

TITOLO DEL PROGETTO		
Management and analysis of molecular and immunological omics data		
ASSEGNO FINANZIATO DA PROGETTO COMPETITIVO <i>(barrare la casella corrispondente)</i>	<input checked="" type="checkbox"/> SI	<input type="checkbox"/> NO
SE IL FINANZIAMENTO È COMPETITIVO L'ENTE FINANZIATORE	AIRC (PI: Prof.ssa Maria Abbondanza Pantaleo)	
PROGETTO/ATTIVITÀ A SCOPO COMMERCIALE <i>(es. sperimentazione profit)</i>	<input type="checkbox"/> SI	<input checked="" type="checkbox"/> NO
CARATTERISTICHE DEL PROGETTO <i>(biomedico/osservazionale/clinico-interventistico/multidisciplinare)</i>	<i>biomedico/osservazionale</i>	
STATO DI APPROVAZIONE DEL PROGETTO DA PARTE DEL COMITATO ETICO <i>(se necessario per il tipo di studio barrare o evidenziare la casella corrispondente)</i>	<input checked="" type="checkbox"/> Ottenuto	<input type="checkbox"/> Da ottenere
DESCRIZIONE DEL PROGETTO <i>(max 800 parole)</i>		



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(1) obiettivi, (2) materiali e metodi, (3) risultati/impatto attesi, (4) attività formativa e (5) di ricerca dell'assegnista

1) Next generation sequencing (NGS) techniques have had an enormous impact in the biomedical field, particularly in the oncology setting, since they have allowed the comprehensive study of the human genome, without the need for prior knowledge and with sustainable times and costs. In particular, techniques such as Whole-Exome Sequencing (WES), RNA-Sequencing (RNA-Seq) and more recently Whole Genome Sequencing (WGS) associated with *ad hoc* bioinformatics analyses have assumed a key role in the molecular study of cancer, both in preclinical and clinical setting. The aim of this project is to analyze NGS data obtained from genomic and transcriptomic sequencing of tumors from patients with visceral sarcomas, which represent a very heterogenous category of rare human tumors, and are therefore frequently orphan of precise characterization of the mutational profile and of specific investments from pharmaceutical companies for drug development.

2) The project will be focused on bioinformatic analysis of NGS data from different categories of visceral sarcomas, that will be obtained by transcriptional, exomic and whole genome profiling of tumor specimens within ongoing studies approved by the local Institutional Ethical Committee of IRCCS Azienda Ospedaliero-Universitaria di Bologna, Italy. The research fellow will develop and use specific bioinformatic pipelines for RNA-seq, WES, and WGS, coupled to functional analyses of the results with specific databases and tools for data annotation and interpretation.

3) The expected results will be to identify specific molecular signatures, driver mutations events, cryptic genomic alterations / rearrangements and specific immunological profiles. The analysis will be focused on different types or different clinical features of visceral sarcomas. The resulting genomic data will be analyzed and compared to clinical outcomes, prognosis and other relevant clinical characteristics.

4/5) The research fellow will be involved in NGS data management and analysis, and in biological interpretation of resulting data. Specific training in NGS library preparation and in data analysis and interpretation is planned. Moreover, the fellow will participate at weekly lab meetings, where the team will discuss all the ongoing projects on visceral sarcomas.

DESCRIZIONE DELLE ATTIVITÀ DELL'ASSEGNISTA

*(per i **nuovi** assegni: max 400 parole; competenze richieste, scansione temporale della formazione, scansione temporale dell'attività, obiettivi primari e secondari)*

*(per i **rinnovi**: max 600 parole – da integrare con la relazione dell'assegnista; formazione raggiunta, attività effettuata, obiettivi raggiunti/competenze acquisite, formazione ancora da acquisire (se pertinente), scansione temporale dell'attività durante il rinnovo)*

Punti

The research fellow is expected to deal intensively with genomic data from visceral sarcomas, including GISTs, intimal sarcomas, leiomyosarcomas, liposarcomas, uterine stromal sarcomas. On these different tumor histotypes, different analyses are planned:

- HRD analysis and mutational profiling of uterine leiomyosarcomas (months 0-3);
- driver rearrangement detection in uterine stromal sarcomas (months 0-4);
- immunological profiling of KIT/PDGFR mutant GIST through gene expression profiling deconvolution (months 2-10);
- mutational and methylation profiling of highly aggressive or non-responder to therapy liposarcomas and leiomyosarcomas (months 4-10);
- WGS, WES, RNA and long read sequencing profiles of quadruple WT GIST, to identify the driver molecular event (months 6-12).



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The fellow will be participating in routine project and troubleshooting discussions, and will be maintaining contacts with collaborators. Moreover, the fellow is expected to present project proceedings at national congresses, and directly take charge of scientific manuscript writing and publication.

Primary objective: Identify molecular signatures and mutational and immunological data relevant in visceral sarcomas.

Secondary objectives: Develop ad hoc bioinformatic pipelines suitable for data analysis in the rare tumor setting.

SE RINNOVO, SI RICORDA DI ALLEGARE ANCHE LA RELAZIONE DELL'ASSEGNISTA CON LA SUA PRODUZIONE SCIENTIFICA.

Scheda attività assistenziale (se prevista)

ATTIVITÀ ASSISTENZIALI DELL'ASSEGNISTA/ N. ORE SETTIMANA (max 18 ore settimanali)
NO
AZIENDA SANITARIA PRESSO CUI SI SVOLGERÀ L'ATTIVITÀ
NA

Si ricorda che, come previsto dagli Accordi sull'impiego nell'attività assistenziale dei Titolari di assegni di ricerca, sottoscritti tra l'Università di Bologna e le Aziende Ospedaliere di riferimento, una volta stipulato il contratto con il vincitore della selezione, il tutor deve consegnare alla Direzione Medica Ospedaliera la relativa modulistica, nella quale andranno riportate le attività qui segnalate.

Management and analysis of molecular and immunological omics data

Research project:

Background and Objectives. Next generation sequencing (NGS) techniques have had an enormous impact in the biomedical field, particularly in the oncology setting, since they have allowed the comprehensive study of the human genome, without the need for prior knowledge and with sustainable times and costs. In particular, techniques such as Whole-Exome Sequencing (WES), RNA-Sequencing (RNA-Seq) and more recently Whole Genome Sequencing (WGS) associated with ad hoc bioinformatics analyses have assumed a key role in the molecular study of cancer, both in preclinical and clinical setting. The aim of this project is to analyze NGS data obtained from genomic and transcriptomic sequencing of tumors from patients with visceral sarcomas, which represent a very heterogeneous category of rare human tumors, and are therefore frequently orphan of precise characterization of the mutational profile and of specific investments from pharmaceutical companies for drug development.

Materials and Methods. The project will be focused on bioinformatic analysis of NGS data from different categories of visceral sarcomas, that will be obtained by transcriptional, exomic and whole genome profiling of tumor specimens within ongoing studies approved by the local Institutional Ethical Committee of IRCCS Azienda Ospedaliero-Universitaria di Bologna, Italy. The research fellow will develop and use specific bioinformatic pipelines for RNA-seq, WES, and WGS, coupled to functional analyses of the results with specific databases and tools for data annotation and interpretation.

Expected Outcomes. The expected results will be to identify specific molecular signatures, driver mutations events, cryptic genomic alterations / rearrangements and specific immunological profiles. The analysis will be focused on different types or different clinical features of visceral sarcomas. The resulting genomic data will be analyzed and compared to clinical outcomes, prognosis and other relevant clinical characteristics.

Research activity and training. The research fellow will be involved in NGS data management and analysis, and in biological interpretation of resulting data. Specific training in NGS library preparation and in data analysis and interpretation is planned. Moreover, the fellow will participate at weekly lab meetings, where the team will discuss all the ongoing projects on visceral sarcomas.

Research activity plan:

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