Discipline MCP5900
Precision Medicine in Cardiology

Concentration area: 5131

Creation: 09/03/2023

Activation: 09/03/2023

Credits: 2

Workload:

Theory (weekly)	Practice (weekly)	Study (weekly)	Duration	Total
3	0	0	10 weeks	30 hours

Professor:

Jose Eduardo Krieger

Objectives:

General: Critical analysis of the main clinical tools and research methodologies in Precision Medicine applied to Cardiology Specific; Indicate and interpret the main genetic tests in patients with hereditary heart diseases, correlating with the clinical phenotype in the main monogenic diseases; classify rare variants found in genetic testing; Conduct a pre-test and post-test counseling consultation for the proband and family, including making a heredogram; Understanding genetic aspects of complex diseases and clinical application of polygenic risk scores; Know and understand innovative diagnostic, risk stratification and therapeutic approaches, using contemporary concepts such as omics, gene editing and cell therapy; Recognize ethical aspects in Precision Medicine. Recognize the need for new social contracts involving public and private service providers, health insurers and employers. Use and storage of data in accordance with current rules, LGPD.

Rationale:

JUSTIFICATION: Proper indication and interpretation of the use of genetic tests, recommended by guidelines, especially international ones, is fundamental for a modern and comprehensive care approach to patients with genetic cardiovascular diseases and for research in this area; The understanding and analysis of scientific methodologies in cardiovascular genetics is important in the training of young researchers, whether in conducting studies that use these methodologies, or for correlation with the line of research of these researchers; The knowledge of innovative concepts that are increasingly being integrated into research and clinical practice is fundamental for the training of avant-garde researchers, so that they are able to assist in the translation of these techniques from the bench to the bedside;

Content:

CONTENT (SUMMARY):General concepts in Precision Medicine in Cardiology (concepts of genetics, genomics and risk assessment); Genetic tests: indications, interpretation, classification of variants and actions; Integration of omics-like biomarkers, images, and laboratory and clinical data to increase diagnostic efficiency and enhance therapeutic accuracy; Genotype x phenotype relationships; Monogenic diseases in Cardiology: cardiomyopathies, arrhythmias, dyslipidemias, vasculopathies and congenital heart diseases;

Complex diseases, GWAS and polygenic risk score; Genetic counseling; New therapeutic approaches: gene therapy and cell therapy

Type of Assessment:

RATING CRITERIA: Attendance, use and participation during classes and discussions; Monography

Bibliography:

Leopold JA, Loscalzo J. Emerging Role of Precision Medicine in Cardiovascular Disease. Circ Res. 2018 Apr 27;122(9):1302-1315. doi: 10.1161/CIRCRESAHA.117.310782. PMID: 29700074; PMCID: PMC6021027. Dainis AM, Ashley EA. Cardiovascular Precision Medicine in the Genomics Era. JACC Basic Transl Sci. 2018 May 30;3(2):313-326. doi: 10.1016/j.jacbts.2018.01.003. PMID: 30062216; PMCID: PMC6059349. Musunuru K, Hershberger RE, Day SM, Klinedinst NJ, Landstrom AP, Parikh VN, Prakash S, Semsarian C, Sturm AC; on behalf of the American Heart Association Council on Genomic and Precision Medicine; Council on Arteriosclerosis, Thrombosis and Vascular Biology; Council on Cardiovascular and Stroke Nursing; and Council on Clinical Cardiology. Genetic testing for inherited cardiovascular diseases: a scientific statement from the American Heart Association. Circ Genom Precis Med. 2020;13:e000067. doi: 10.1161/HCG.0000000000000067 Wilde AAM, Semsarian C, Márquez MF, Sepehri Shamloo A, Ackerman MJ, Ashley EA, Sternick EB, Barajas-Martinez H, Behr ER, Bezzina CR, Breckpot J, Charron P, Chockalingam P, Crotti L, Gollob MH, Lubitz S, Makita N, Ohno S, Ortiz-Genga M, Sacilotto L, Schulze-Bahr E, Shimizu W, Sotoodehnia N, Tadros R, Ware JS, Winlaw DS, Kaufman ES; Document Reviewers, Aiba T, Bollmann A, Choi JI, Dalal A, Darrieux F, Giudicessi J, Guerchicoff M, Hong K, Krahn AD, MacIntyre C, Mackall JA, Mont L, Napolitano C, Ochoa JP, Peichl P, Pereira AC, Schwartz PJ, Skinner J, Stellbrink C, Tfelt-Hansen J, Deneke T. European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the State of Genetic Testing for Cardiac Diseases. Heart Rhythm. 2022 Jul;19(7):e1-e60. doi: 10.1016/j.hrthm.2022.03.1225. Epub 2022 Apr 4. PMID: 35390533. O'Sullivan JW, Raghavan S, Marquez-Luna C, Luzum JA, Damrauer SM, Ashley EA, O'Donnell CJ, Willer CJ, Natarajan P; on behalf of the American Heart Association Council on Genomic and Precision Medicine; Council on Clinical Cardiology; Council on Arteriosclerosis, Thrombosis and Vascular Biology; Council on Cardiovascular Radiology and Intervention; Council on Lifestyle and Cardiometabolic Health; and Council on Peripheral Vascular Disease. Polygenic risk scores for cardiovascular disease: a scientific statement from the American Heart Association. Circulation. 2022;146:e93–e118. doi: 10.1161/CIR.0000000000001077 Richards S, Aziz N, Bale S, Bick D, Das S, Gastier-Foster J, Grody WW, Hegde M, Lyon E, Spector E, Voelkerding K, Rehm HL; ACMG Laboratory Quality Assurance Committee. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med. 2015 May;17(5):405-24. doi: 10.1038/gim.2015.30. Epub 2015 Mar 5. PMID: 25741868; PMCID: PMC4544753. Pina-Neto JM. Genetic counseling. J Pediatr (Rio J). 2008;84(4 Suppl):S20-26. doi:10.2223/JPED.1782 Mengying Dong, Jiangen Liu, Caixia Liu, He Wang, Wei Sun, Bin Liu; CRISPR/CAS9: A promising approach for the research and treatment of cardiovascular diseases, Pharmacological Research, Volume 185, 2022, 106480, ISSN 1043-6618, https://doi.org/10.1016/j.phrs.2022.106480 . Sadek H, Olson EN. Toward the Goal of Human Heart Regeneration. Cell Stem Cell. 2020 Jan 2;26(1):7-16. doi: 10.1016/j.stem.2019.12.004. PMID: 31901252; PMCID: PMC7257208.

Languages taught:

Portuguese

Class type:

Não-Presencial

Additional class type information:

A porcentagem da disciplina que ocorrerá no sistema não presencial (1- 100%): - 87,5% (7 das 8 aulas); Detalhamento das atividades que serão presenciais e das que serão desenvolvidas via remota, com discriminação do tempo de atividade contínua online: - Presenciais - atividades práticas: sequenciamento, classificação de variantes, elaboração de laudos/relatórios, aconselhamento genético - Remota ou presencial (a critério do aluno): aulas expositivas, discussão de artigos, discussão de casos clínicos; Especificação se as aulas, quando online, serão síncronas ou assíncronas: - Síncronas, sempre; Descrição do tipo de material e/ou conteúdo que será disponibilizado para o aluno e a plataforma que será utilizada: - Disponibilizados artigos científicos que serão discutidos; plataforma ZOOM; Definição sobre a presença na Universidade e, quando necessária, discriminar quem deverá estar presente (professora/professor; aluna/aluno; ambos): - Presença obrigatória na aula 8 de professores e alunos; Descrição dos tipos e da frequência de interação entre aluna/aluno e professora/professor (somente durante as aulas; fora do período das aulas; horários; por chat/e-mail/fóruns ou outro). - Durante o período das aulas: via ZOOM; Fora do período das aulas: via e- mail; A forma de controle da frequência nas aulas: - Nome na ferramenta ZOOM ou lista de presença; Informação sobre a obrigatoriedade ou não de disponibilidade de câmera e áudio (microfone) por parte dos alunos: - Obrigatório câmera e microfone; A forma de avaliação da aprendizagem (presencial/remota). - Avaliação remota