Cardiomiopatie Unit

Descrizione

Responsabile *in via di nomina*

Contatti
cardiomiopatie@aou-careggi.toscana.it
Tel. 055 794 5138 dal lunedì al venerdì ore 7:30 – 08:30, 13:00 – 14:00

Le cardiomiopatie sono un'entità clinica eterogenea, spesso misconosciuta e quindi non diagnosticata, ma importante dal punto di vista epidemiologico e complessa da punto di vista assistenziale, con aspetti culturali peculiari, la cui gestione necessita di competenze specifiche ed esperienza nel settore. Queste le patologie trattate:

- cardiomiopatie genetiche, dell'adulto e pediatriche: cardiomiopatia ipertrofica, cardiomiopatie dilatative, cardiomiopatia aritmogena, cardiomiopatie restrittive, cardiomiopatie associate a canalopatie, cardiomiopatie non classificate.
- morte cardiaca improvvisa giovanile e relativo inquadramento diagnostico e terapeutico delle famiglie colpite
- cardiomiopatie secondarie (tossiche e da trattamenti oncologici, infettive, infiltrative, autoimmuni, ecc.)
- Sindrome di Marfan e patologie correlate, afferenti al Centro di Riferimento regionale Diagnosi e trattamento della sindrome di Marfan di cui questa Unit svolge la valutazione cardiologica

Funzioni

- gestione del percorso di diagnosi e cura dei pazienti affetto da cardiomiopatia, in contesto multidisciplinare e multiprofessionale, in rete con altre strutture, intraospedaliere ed inter-ospedaliare (ad esempio col Prof Iacopo Olivotto – AOU Meyer per valutazione congiunta delle cardiomiopatie pediatriche e metaboliche, AOU Senese per la gestione trapiantologica etc) e territoriale (Medicina Generale, specialista territoriale, ecc.)
- organizzazione di consulenza clinico – genetica per le cardiomiopatie a carattere familiare, e eventuali indagini genetiche mirate, in collaborazione con la SOD Diagnostica Genetica
- promozione della ricerca scientifica, applicazione e sviluppo di protocolli e Linee guida
- sviluppo di progetti specifici nell'ambito della didattica per studenti del corso di laurea, per medici in formazione specialistica e fellow, organizzazione eventi scientifici e formativi di livello locale, nazionale ed internazionale
- sensibilizzazione e informazione sul tema delle malattie cardiologiche rare, in collaborazione con le associazioni di pazienti, la Fondazione Careggi ed altri Enti di supporto

Percorso di diagnosi e cura

- prima visita specialistica cardiologica per miocardiopatia (presa in carico del paziente presso Ambulatorio cardiovascolare Clinica Medica secondo piano)
- prescrizione di indagini strumentali e di laboratorio
- programmazione interventi terapeutici (farmacologici o chirurgici)
- programmazione interventi di riabilitazione
- follow-up

Modalità di accesso alle prestazioni

Contattare il Centro (preferibilmente per e-mail) indicando il sospetto diagnostico o la patologia accertata; il paziente sarà successivamente ricontattato per concordare la data dell'appuntamento.

Collaborazioni

- Cardioanestesia
- Cardiochirurgia
- Diagnostica cardiovascolare
- Diagnostica Genetica
- Medicina nucleare
- Riabilitazione cardiologica
- Cardiologia Pediatrica e Studio Malattie Metaboliche e Muscolari Ereditarie -Azienda Ospedaliero-Universitaria Meyer

Collaborazioni internazionali

Il Centro collabora attivamente con i principali centri universitari italiani che si occupano di cardiomiopatie, e con vari numerosi gruppi di ricerca:

- Dipartimento di Fisiologia dell'Università degli studi di Firenze
- Dipartimento di Farmacologia dell'Università degli studi di Firenze
- HCM Center, Tufts Medical Center (Boston, USA)
- HCM Center, Minneapolis Heart Institute (Minneapolis, USA)
- Magdi Yacoub Research Network

Progetti di ricerca

- Progetti finanziati dal Ministero dell'Università e della Ricerca Scientifica e Tecnologica:

-COFIN 2002 e 2006 su "Determinanti clinici dello scompenso cardiaco e della morte improvvisa nei pazienti con cardiomiopatia ipertrofica con mutazioni sarcomeriche";

- Unione Europea: Collaborative project FP7-HEALTH-2009-single-stage: "Bench-to-bedside InteGrated approach to familial hypertrophic cardiomyopathy: to the HEART of the disease (BIG-HEART)".

- Ricerca Finalizzata del Ministero della Salute:

-RF 2010 – 2313451 “Hypertrophic cardiomyopathy: new insights from deep sequencing and psychosocial evaluation”.

-NET-2011-02347173 (Mechanisms and treatment of coronary microvascular dysfunction in patients with genetic or secondary left ventricular hypertrophy)

-RF-2013-02356787 (Left ventricular hypertrophy in aortic valve disease and hypertrophic cardiomyopathy: genetic basis, biophysical correlates and viral therapy models)

- Telethon Italia (GGP13162)

Studi clinici in corso

- Centro Coordinatore dello studio Liberty-HCM. “A Phase 2/3, Pivotal, Randomized, Double-Blind, Placebo-Controlled Study to Evaluate the Effect of GS 6615 on Exercise Capacity in Subjects with Symptomatic Hypertrophic Cardiomyopathy”.
- NIH Grant U01HL117006-01A1; HCMR Protocol “HCMR – Novel Predictors of Outcome in Hypertrophic Cardiomyopathy”. Inizio arruolamento previsto in autunno. Dr.Olivotto, investigator. Sponsor: US National Institutes of Health.
- Centro coordinatore dello studio “Ranolazine in patients with symptomatic hypertrophic cardiomyopathy: a pilot study assessing the effects on exercise capacity, diastolic function and symptomatic status. A Randomized, Double blind, Placebo controlled, Parallel Group, Pilot Study, in patients with Symptomatic Hypertrophic Cardiomyopathy.
- Centro partecipante allo studio: “ATTRACT Study (AT1001 Therapy Compared to Enzyme Replacement in Fabry Patients with AT1001-responsive Mutations: a Global Clinical Trial)”

Registri internazionali

- SHaRe: The Sarcomeric Human Cardiomyopathies Registry. Brigham and Women’s Hospital; Children’s Hospital Boston; Stanford University; University of Michigan; the Florence Centre for Cardiomyopathies (IT); University College, London (UK); and Erasmus Medical Center (Rotterdam, NL). www.thesharegistry.com
- Fabry Registry: A Multi-Center, International, Longitudinal, Observational Program Designed to Track the Natural History and Outcomes of Patients with Gaucher, Fabry, MPS I, and Pompe Disease. – codice DIREGC07006
- EuroObservational Research Program (EORP): Cardiomyopathy Pilot Registry. European Society of Cardiology

Pubblicazioni

Queste le pubblicazioni scientifiche degli ultimi anni a cui hanno partecipato i medici dell'equipe della Cardiomiopatie Unit

- Quarta G, Papadakis M, Donna PD, Maurizi N, Iacovoni A, Gavazzi A, Senni M, Olivotto I. Grey zones in cardiomyopathies: defining boundaries between genetic and iatrogenic disease. Nat Rev Cardiol. 2017 Feb;14(2):102-112. doi: 10.1038/nrcardio.2016.175. Review. PubMed PMID: 27830773.
- Picano E, Ciampi Q, Citro R, D'Andrea A, Scali MC, Cortigiani L, Olivotto I, Mori F, Galderisi M, Costantino MF, Pratali L, Di Salvo G, Bossone E, Ferrara F, Gargani L, Rigo F, Gaibazzi N, Limongelli G, Pacileo G, Andreassi MG, Pinamonti B, Massa L, Torres MA, Miglioranza MH, Daros CB, de Castro E Silva Pretto JL, Beleslin B, Djordjevic-Dikic A, Varga A, Palinkas A, Agoston G, Gregori D, Trambaiolo P, Severino S, Arystan A, Paterni M, Carpeggiani C, Colonna P. Stress echo 2020: the international stress echo study in ischemic and non-ischemic heart disease. Cardiovasc Ultrasound. 2017 Jan 18;15(1):3. doi: 10.1186/s12947-016-0092-1. PubMed PMID: 28100277; PubMed Central PMCID: PMC5242057.
- Maurizi N, Tanini I, Olivotto I, Amendola E, Limongelli G, Losi MA, Allocca G, Perego GB, Pieragnoli P, Ricciardi G, De Filippo P, Ferrari P, Quarta G, Viani S, Rapacciuolo A, Bongiorni MG, Cecchi F. Effectiveness of subcutaneous implantable cardioverter-defibrillator testing in patients with hypertrophic cardiomyopathy. Int J Cardiol. 2017 Jan 4. pii: S0167 5273(16)33706-8. doi: 10.1016/j.ijcard.2016.12.187. [Epub ahead of print] PubMed PMID: 28073660.
- Sciagrà R, Calabretta R, Cipollini F, Passeri A, Castello A, Cecchi F, Olivotto I, Pupi A. Myocardial blood flow and left ventricular functional reserve in hypertrophic cardiomyopathy: a (13)NH(3) gated PET study. Eur J Nucl Med Mol Imaging. 2017 Jan 3. doi: 10.1007/s00259-016-3603-2. [Epub ahead of print] PubMed PMID: 28050630.
- Re F, Zachara E, Avella A, Baratta P, di Mauro M, Uguccioni M, Olivotto I. Dissecting functional impairment in hypertrophic cardiomyopathy by dynamic assessment of diastolic reserve and outflow obstruction: A combined cardiopulmonary-echocardiographic study. Int J Cardiol. 2017 Jan 15;227:743-750. doi: 10.1016/j.ijcard.2016.10.067. PubMed PMID: 27839814.
- Tomberli B, Cappelli F, Perfetto F, Olivotto I. Abrupt Onset of Refractory Heart Failure Associated With Light-Chain Amyloidosis in Hypertrophic Cardiomyopathy. JAMA Cardiol. 2017 Jan 1;2(1):94-97. doi: 10.1001/jamacardio.2016.3894. PubMed PMID: 27806176.
- Castagnoli H, Ferrantini C, Coppini R, Passeri A, Baldini K, Berti V, Cecchi F, Olivotto I, Sciagrà R. Role of quantitative myocardial positron emission tomography for risk stratification in patients with hypertrophic cardiomyopathy: a 2016 reappraisal. Eur J Nucl Med Mol Imaging. 2016 Dec;43(13):2413-2422. PubMed PMID: 27527796.
- Barlocco F, Olivotto I. Can anthropology improve our care of inherited cardiac arrhythmias? A modest proposal. Heart Rhythm. 2016 Dec;13(12):2395-2398. doi: 10.1016/j.hrthm.2016.08.042. PubMed PMID: 27590433.
- Hughes DA, Nicholls K, Shankar SP, Sunder-Plassmann G, Koeller D, Nedd K, Vockley G, Hamazaki T, Lachmann R, Ohashi T, Olivotto I, Sakai N, Deegan P, Dimmock D, Eyskens F, Germain DP, Goker-Alpan O, Hachulla E, Jovanovic A, Lourenco CM, Narita I, Thomas M, Wilcox WR, Bichet DG, Schiffmann R, Ludington E,Viereck C, Kirk J, Yu J, Johnson F, Boudes P, Benjamin ER, Lockhart DJ, Barlow C, Skuban N, Castelli JP, Barth J, Feldt-Rasmussen U. Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study. J Med Genet. 2016 Nov 10. pii: jmedgenet-2016-104178. doi: 10.1136/jmedgenet-2016-104178. [Epub ahead of print] PubMed PMID: 27834756.

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- Giovannelli F, Simoni D, Gavazzi G, Giganti F, Olivotto I, Cincotta M, Pratesi A, Baldasseroni S, Viggiano MP. Electrophysiological correlates of word recognition memory process in patients with ischemic left ventricular dysfunction. Clin Neurophysiol. 2016 Sep;127(9):3007-13. doi: 10.1016/j.clinph.2016.06.022. PubMed PMID: 27469528.
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- Krishnamoorthy N, Gajendrarao P, Olivotto I, Yacoub M. Impact of disease-causing mutations on inter-domain interactions in cMyBP-C: a steered molecular dynamics study. J Biomol Struct Dyn. 2016 Jun 30:1-7. [Epub ahead of print] PubMed PMID: 27267291.
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Data
13/04/2026