

## **Preliminary Program**

## Wednesday, April 27

| 8.00 p.m.               | *****Welcome dinner***** @Cenobio Dei Dogi Hotel, La Playa Restaurant |
|-------------------------|---|
| Robert Hofstra memorial | Maria Alves, Isabella Ceccherini, Nikhil Thapar                       |
| POIC e dintorni welcome | Alessandro Palmitelli   |

## Thursday, April 28

| Thursday, April 20                          |  |  |
|---|--|--|
| 8.45 a.m.                                   | Session1: <b>Symptoms and diagnosis</b>                                      |  |
|   | Chairpersons: <b>Maria Judit Molnar &amp; Carlo Di Lorenzo</b>               |  |
| Paolo Gandullia                             | Clinical description   |  |
| Osvaldo Borrelli                            | Diagnostic methods   |  |
| Alessio Pini Prato                          | The role of a surgery in the differential diagnosis                          |  |
| 9.45 a.m.                                   | Session2: <b>Genetics and pathogenetic mechanisms</b>                        |  |
|   | Chairpersons: <b>Isabella Ceccherini &amp; Federica Viti</b>                 |  |
| Isabella Ceccherini                         | Genetics of the visceral myopathy: a focus on the ACTG2 gene                 |  |
| Robert Heuckeroth                           | Hypothesis on pathogenetic mechanisms of VSCM                                |  |
| 10.25 *****Coffee break***** (25')          |  |  |
| Maria Alves                                 | Understanding the genetic etiology of CIPO: one gene at a time               |  |
| Elena Bonora                                | Experience on mitochondrial genes involved in VSCM                           |  |
| Discussion on diagnosis a                   | nd genetics (20')  |  |
| 11.50 a.m.                                  | Session3: <b>Models and endpoints</b>  |  |
|   | Chairpersons: <b>Maria Alves &amp; Massimo Vassalli</b>                      |  |
| Sohaib Hashmi                               | New models for visceral myopathy: Applying lessons from studying ACTG2 R257C |  |
|   | in human intestinal SMCs   |  |
| Federica Viti                               | Morpho-mechanical phenotyping of fibroblasts from VSCM patients              |  |
| Arti Ahluwalia                              | Advanced intestinal models   |  |
| 12.50 a.m 2.15 p.m. *****Lunch***** (1h25') |  |  |
| Antonio Galeone                             | ACTG2 involvement in Drosophila  |  |
| Pascal de Santa Barbara                     | Phenotypic switch of SMC in CIPO patients                                    |  |
| Maxime Mahe                                 | Modelling human gut development using iPSC                                   |  |
| Silvia Perin                                | Development of enteric smooth muscle   |  |
| Discussion on models and                    | l endpoints (20')  |  |
| 3.55 p.m. *****Coffee break***** (25')      |  |  |
| 4.20 p.m.                                   | Session4: Current management and futuristic treatments                       |  |
|   | Chairpersons: Nikhil Thapar & Roberto De Giorgio                             |  |
| Nikhil Thapar                               | Current avenues of management in paediatric and adult VSCM                   |  |
| Roberto De Giorgio                          | Pharmacotherapy and manipulation of microbiota                               |  |
| Antonella Diamanti                          | Diet and Parenteral Nutrition  |  |

| Luigi  | Dall'Oglio   | and                          | Surgery and Intestinal Transplant      |
|--|--------------|------------------------------|--|
| Floren   | ice Lacaille |                              |  |
| Paolo  | De Coppi     |                              | Regenerative Medicine (remote session) |
| Discussion on VSCM management and treatments (20') |              | agement and treatments (20') |  |

| 0.00        | ****            |
|-------------|-----------------|
| 8.00 p.m.   | *****Dinner**** |
| 0.00 p.111. | Billinei        |

## Friday, April 29

| 8.45 a.m.            | Session5: <b>Research support strategies</b>                           |
|----------------------|--|
|                      | Chairpersons: Alessandro Palmitelli & Carlotta De Filippo              |
| Antonio Contessa and | Families' perspectives on living with visceral myopathy                |
| Laurence Lancon      |  |
| Carlotta De Filippo  | Citizen Science  |
| Chiara Baldo         | Biobanks: sharing human biological material for rare disease research  |
| Ruggero Di Maulo     | Pathology registries for patient associations: opportunity and threats |
| Salvo Leone          | Patients International Alliances: How to get there                     |
| 10.45 a.m. *****Coff | fee break**** (25')  |
| 11.10 a.m.           | Session6: <b>Round table</b>   |
|                      | Chairpersons: <b>Carlo di Lorenzo, Carlotta De Filippo</b>             |
| 12.45 a.m. – 2 p.m.  | *****Lunch****   |