First meeting on Italian research about alpha-1 antitrypsin deficiency

The Italian association of patients of alpha-1 antitrypsin deficiency, Associazione a1AT onlus, has organised and hosted the first meeting of Italian researchers and clinicians devoted to alpha-1 antitrypsin deficiency. On February 25th, the beautiful city of Brescia has received the participants from all over Italy for a full day dedicated to both basic research and clinical aspects of this disease, with the main objectives of sharing data and opinions, establishing and reinforcing collaborative efforts.

The morning session, moderated by Ilaria Ferrarotti, Anna Fra and Elena Miranda, was dedicated to basic research. The first speaker, Ilaria Ferrarotti, presented an update on the epidemiology of alpha-1 antitrypsin deficiency in Italy. Next, Elena Miranda talked about the nature of pathological antitrypsin polymers and how monoclonal antibodies can be used to study them. In the third presentation Anna Fra illustrated an in silico approach to predict the pathogenicity of point mutations, as well as the recent finding of an unusual mutant variant that forms polymers with a different nature to the Z ones. This was followed by a biophysical look into how to study serpin polymers by Mauro Manno. The session continued with two talks by Federica Michielin and Riccardo Ronzoni about the use of induced pluripotent stem cells differentiated to hepatocytes as a model system to study alpha-1 antitrypsin deficiency. In the following double act, Nicola Brunetti-Pierri and Pasquale Piccolo presented their research on the cellular effects of Z antitrypsin in the liver of transgenic PiZ mice, reporting the activation of the JNK signalling pathway and the regulatory role of specific miRNAs in this model system. The last speaker of the morning session, Francesco Cappello, nicely illustrated the anatomy of the liver and the role of chaperones in protein conformational diseases.

At the end of the morning session, packed with first level research and discussions during the question times, everybody was happy to stay sit for the key lecture delivered by Prof. Francesco Callea. With the authority of whom has long studied the alpha-1 antitrypsin deficiency, Prof. Callea shared with the audience his knowledge and views about how the antitrypsin polymers remain trapped and cause liver disease.

The afternoon session was moderated by Claudio Tantucci and Angelo Corsico, and dedicated to different clinical aspects of alpha-1 antitrypsin deficiency, including uncommon manifestation of the disease. The first speech was made by Stefano Aliberti, who held a marvellous lecture about bronchiectasis and discussed with the audience about the relationship between bronchiectasis and alpha1 antitrypsin deficiency. Bruno Balbi illustrated the microbioma project, aiming at a definition of the airway bacterial and viral microbioma in patients with apha-1 antitrypsin deficiency, either on augmentation therapy or not, compared with non- apha-1 antitrypsin deficiency-chronic obstructive pulmonary disease patients. Laura Pini presented the results of a clinical research purposed to better understand the possible association between alfa1-antitrypsin deficiency and vascular diseases. Later on, Gina Gregorini, with her great expertise, gave a review about vasculitis, and Maurizio Fuoti nicely illustrated the characteristics of the disease and the potential approach to new therapies during childhood. At the end of the session Luciano Corda, with his great knowledge about the disease, and Federica Benini briefly discussed with the audience about some critical issues that could be developed in next clinical studies.

The meeting was closed by Nuccia Gatta, the soul of the Italian alpha-1 patient association, who remarked again the high level of the scientific and clinical speakers and encouraged the active community of researchers and doctors dealing with alpha-1 antitryspin deficiency to continue their efforts in order to understand, mitigate and hopefully cure this condition