Since January 2020 Elsevier has created a COVID-19 resource centre with free information in English and Mandarin on the novel coronavirus COVID-19. The COVID-19 resource centre is hosted on Elsevier Connect, the company's public news and information website.

open his eyes on command. His clinical condition then deteriorated over the following 3 days with worsening oxygen saturations and blood gases. He became unresponsive despite sedation being stopped. CT head was normal but a repeat CT chest showed worsening left lower lobe necrosis. After a discussion with the family it was agreed additional treatment would be futile and care should be withdrawn. He died peacefully 20 days after admission to hospital, 22 days after testing positive for COVID-19.
Conclusion: To our knowledge, this is the first reported case of necrotising pneumonia (NP) associated with COVID-19 in an individual with CF and the first associated with *Nocardia* infection. We hope by reporting this case it will highlight the continued risks of COVID-19 infection relevant to individuals with CF of any age.

WS21 – Potpourri of genetics and biomarkers

WS21.01
AQP5 and *CFTR*, two genes associated with pseudo-aquagenic palmoplantar keratoderma?

E. Girodon[1], M. Lopez[1], B. Sperelakis-Beedham[1], E. Bourrat[2], N. Gaitch[1], F. Houriez[1], B. Martinez[1], I. Fajac[3], P.-R. Burgel[4], G. Hickman[2], M.-P. Audrézet[5], A.-M. Guerrot[6], F. Cabet[7], M. Gerfaud-Valentin[8], R. Nove-Josserand[9], C. Raynal[10], A. Pagin[11], M.-P. Reboul[12], A. De Becdelièvre[13], I. Callebaut[14], T. Bienvenu[1]. *[1]APHP.Centre-Université de Paris, Hôpital Cochin, Service de Médecine Génomique des Maladies de Système et d'Organe, Paris, France; [2]CRMR MAGEC Nord St Louis, Hôpital-Saint Louis, Service de Dermatologie, Paris, France; [3]APHP.Centre-Université de Paris, Hôpital Cochin, Physiologie-Explorations Fonctionnelles, Paris, France; [4]APHP.Centre-Université de Paris, Hôpital Cochin, Service de Pneumologie, CRCM, Paris, France; [5]CHRU de Brest, Laboratoire de Génétique Moléculaire, Brest, France; [6]CHU de Rouen, Service de Génétique, Rouen, France; [7]Hospices Civils de Lyon, Groupement Hospitalier Est, CBPE, Service Biochimie Biologie Moléculaire Grand Est, Lyon, France; [8]Hospices Civils de Lyon, Hôpital de la Croix Rousse, Service de Médecine Interne, Lyon, France; [9]Centre Hospitalier Lyon Sud, Service de Médecine Interne, CRCM, Pierre-Benite, France; [10]CHU de Montpellier, PhyMedExp, INSERM U1046, Laboratoire de Génétique Moléculaire, Montpellier, France; [11]CHRU de Lille, Service de Toxicologie et Génopathies, Lille, France; [12]Groupe Hospitalier Pellegrin, C.H.U. de Bordeaux, Laboratoire de Génétique Moléculaire, Bordeaux, France; [13]AP-HP. Hôpitaux Universitaires Henri-Mondor, Hôpital Henri Mondor, Laboratoire de Génétique, Créteil, France; [14]IMPMC, Sorbonne Université, Muséum National d'Histoire Naturelle, UMR7590 CNRS, Paris, France*

Objectives: Aquagenic palmoplantar keratoderma (APPK) is frequently observed in patients with CF but it also occurs sporadically, often in young women. An excess of *CFTR* gene variant carriers has been observed in patients and a CFTR-related disorder (CFTR-RD) could sometimes be unmasked. However, the majority of cases being unsolved, other genes might be at play, such as *AQP5*, the aquaporine 5 encoding gene, involved in Bothnian-type palmoplantar keratoderma (PPK), which is a permanent PPK worsened by water immersion.
Methods: The coding regions of *CFTR* and *AQP5* genes were studied in 187 patients enroled for isolated APPK, using next-generation sequencing (AmpliSeq for Illumina). Clinical and familial data were collected, along with sweat test results. Pathogenicity of *CFTR* and *AQP5* variants was assessed according to international recommendations.
Results: 163/187 (87.2%) of patients were women. Median age was 22 years [10–41]. Minor respiratory symptoms were reported in 16% of cases. The sweat test results, available for 29 patients, was either positive (n = 1), intermediate (n = 13) or negative (n = 14). One patient was diagnosed with CF (0.5%), 12 with a CFTR-RD (6.4%) along with 27 identified as CF carriers (7.2%). Six patients were found to carry an *AQP5* variant, of which 5 were missense in the C-terminal domain. Two of the 6 patients also carried a *CFTR* variant. Three had discrete PPK apart from water immersion. A moderate impact of these *AQP5* variants was suggested by 3D structure modeling.
Conclusion: The study confirms the role of CFTR in APPK. The role of AQP5 is also suggested, especially C-terminal variants which could mimic an APPK instead of a Bothnian-type PPK. This prompts searching for permanent PPK, even if discrete, as well as other cases of PPK in the patients' families plusstudying *AQP5* alongside *CFTR*. The study raises the hypothesis of a multifactorial etiology, with the possible interplay of *CFTR* and *AQP5* variants.

WS21.02
Cystic fibrosis modifier genes and bacterial infections in Spanish patients

E. Granizo-Rodriguez[1], M. Baeta[1], T. Kleinbielen[1], O. Padura[2], P. Arizti[3], F. Baranda[4], M.M. De PANCORBO[1]. *[1]University of the Basque Country UPV/EHU, BIOMICs Research Group, Vitoria-Gasteiz, Spain; [2]Asociación de Fibrosis de Euskadi, Donostia - San Sebastián, Spain; [3]Asociación Balear de Fibrosis Quística, Palma, Spain; [4]Hospital de Cruces, Barakaldo, Spain*

Background: Being aware of the wide diversity of cystic fibrosis (CF) mutations, it is not surprising the extensive range of phenotypic diversity between CF patients. Due to this diversity, modifier genes have been described to play a major role on bacterial infection, inflammation or even pulmonary damage/remodeling. Owing to this capacity to modulate the severity of the disease, the study of the modifier genes could be a useful tool to dig deep on the pathogeny as well as to identify new therapeutic targets.
Objective: Thirty-three SNPs corresponding to 19 modifier genes were analysed on 441 individuals to assess the potential risk or protection against bacterial infection caused by *S.aureus*, *P. aeruginosa*, *B. cepacia*, *Non-tuberculous bacteria* or *S. maltophilia*.
Methods: Three hundred five saliva swab samples were collected from 109 CF patients and their parents from Spanish CF associations. A further 136 DNA samples from healthy donors were collected as controls. Additional information concerning clinical and lifestyle data was given by the participants. DNA genotyping was performed via Iplex Gold (Agena Bioscience) or via SnapShot, depending on the complexity of the target. Statistical analysis were performed.
Results: Preliminary results seem to indicate that several SNPs from different genes are significantly associated with 1 or even more risk genetics models, as do haplotypes. After processing the SNPs, both as single markers and as combined haplotypes, we were able to establish a correlation of certain alleles with the infection by *S. aureus* and *S. maltophilia*, not only in genotype models but also in haplotype ones.
Conclusion: According to the results, certain modifier genes studied herein seem to influence on infectious process in CF disease in Spanish patients. Further studies are needed to validate the results in a large sample set.

WS21.03
Low-cost chain termination DNA sequencing PCR reaction to diagnose *CFTR* gene mutations

M.U. Ghani[1], M.F. Sabar[1], F.I. Awan[1]. *[1]University of the Punjab, Centre for Applied Molecular Biology, Lahore, Pakistan*

Objectives: Cystic fibrosis (CF) is a life limiting disease caused by autosomal genomic mutations in *CFTR* gene. CF is common in developed countries but not uncommon in developing/low income countries although it is under-diagnosed in those countries due to lack of diagnostic facilities and financial constraints. As no genomic mutations-based public diagnostics facility is available in Pakistan for CF patients, a low-cost assay to diagnose CF causative genomic mutations in Pakistani patients is proposed.
Methods: Capillary-based DNA Sanger sequencing method to analyse DNA fragments of about 600 bp sequence in one reaction was targeted. The quantity of "terminator ready reaction premix" (2.5X, BigDye™ Terminator v3.1) was gradually reduced from 4 ul to 01 µl and the quantities of the remaining reagents of chain termination sequencing PCR were optimised stoichiometrically.
Results: The modified chain termination assay successfully analysed more than 50 DNA samples without effecting the mutation detection capability and quality of DNA sequencing electropherogram generated on ABI-3130XL Genetic Analyzer.