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Title: Prevalence of alpha-1 antitrypsin deficiency (AATD) and frequencies of alleles PI*S and PI*Z in patients with COPD in Brazil

Dr. Rodrigo 22312 Russo rodrigo_russo@yahoo.com.br MD [1], Mrs. Laura 22313 Zillmer lahzillmer@hotmail.com [1], Dr. Oliver 22314 Nascimento olivernascimento@yahoo.com.br MD [1], Dr. Gildo Francisco 22315 Santos, Junior gildo.junior@afip.com.br [4], Dr. Leandro 22328 Fritscher (leandro.fritscher@pucrs.br MD [2], Dr. Marc 26809 Miravitlles marcm@clinic.ub.es MD [3], Dr. Fernando 26853 Lundgren lundgrenf@gmail.com MD [5], Mrs. Marcela Amorim 26861 Alves m.amorimalves@gmail.com MD [5], Mrs. Heicilany Del Carlos 26870 Gondim heici@uol.com.br MD [7], Dr. Maria Penha Uchoa 26875 Sales puchoasales152@gmail.com MD [6], Mrs. Maria Vera Cruz 26916 Oliveira maria_vera@uol.com.br MD [8], Dr. Altay A.L. 27101 Souza altayals@gmail.com [1] and Prof. Dr José Roberto 27111 Jardim jardimpneumo@gmail.com MD [1]. [1] Pulmonary Reabilitation Center/Association for Assistance To Disabled Children (AACD), Federal University of São Paulo, São Paulo, SP, Brazil ; [2] Department of Pulmonary Medicine, Pontifical Catholic University of Rio Gande Do Sul, Porto Alegre, SP, Brazil ; [3] Department of Pneumology, Vall D'Hebron Hospital, Barcelona, Spain ; [4] Division of Molecular Biology, AFIP (Association Research Incentive Fund), São Paulo, SP, Brazil ; [5] Department of Pulmonary Medicine, Otávio De Freitas Hospital, Recife, PE, Brazil ; [6] Department of Pulmonary Medicine, Messejana Hospital, Fortaleza, CE, Brazil ; [7] General Hospital Dr Alberto Rassi, Department of Pulmonary Medicine, Goiania, GO, Brazil and [8] Department of Pulmonary Medicine, Public Employee Hospital of São Paulo State, São Paulo, SP, Brazil .

Body: Introduction: AATD affects about one in 2000–5000 individuals. According to PLATINO study, there are 5 to 7 million patients with COPD in Brazil, however there is no epidemiological data on the prevalence of AATD or the frequency of occurrence of deficiency alleles among them. Objective: to estimate the prevalence of AATD in patients with COPD in Brazil and the frequency of occurrence of deficiency alleles. Methods: dried blood spot (DBS) samples were tested for ATT in 1019 patients with COPD in five Brazilian cities. Eluated was prepared from DBS 6mm diameter perforated discs (Whatman™, GE, 903, lot W101, USA) and 200 µmL phosphate-buffered saline (pH 7.4) overnight at 4° C. All individuals with DBS dosage less than 2.64 mg/dl (upper limit of the confidence interval of the cutoff point) were considered as possible disabled and an AAT serum dosage was performed. All individuals with a serum dosage <113 mg/dl had a genotyping performed. Serum and DBS samples were determined using the rate immune nephelometric method (Siemens, BNII). Results: 99 patients had a DBS dosage equal or less than 2.64 mg/dl and 26 of them a serum dosage <113 mg/dl. The prevalence of AATD in patients with COPD was 2.55%. Frequency of the main deficiency phenotypes were: PiMZ (1.17%), PiZZ (0.78%), PiMS (0.40%), PiSS (0.1%), and

PiSZ (0.1%). Conclusion: this is the first study designed to establish the prevalence of AATD and frequency of deficiency alleles in patients with COPD in Brazil. The prevalence of 2.55% reflects the rates found in studies around the world and reinforces the need for screening all patients with COPD followed by genotyping patients with AATD.