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Cystic Fibrosis Research News

Title:
Clinical outcomes of a large cohort of individuals with the F508del/5T;TG12 CFTR genotype

Lay Title:
patients with F508del/5T;TG12 CFTR genotype

Authors:
Antonella Tosco[a], Alice Castaldo[a], Carla Colombo[b], Laura Claut[b], Vincenzo Carnovale[c], Paola Iacotucci[c], Marco Lucarelli[d,e], Giuseppe Cimino[f], Benedetta Fabrizzi[g], Nicole Caporelli[g], Fabio Majo[h], Fabiana Ciciriello[h], Rita Padoan[i], Piercarlo Poli[i], Giovanni Taccetti[l], Claudia Centrone[m], Rosaria Casciaro[n], Carlo Castellani[n], Donatello Salvatore[o], Carmela Colangelo[o], Paolo Bonomi[p], Giuseppe Castaldo[q,r], Vito Terlizzi[l]

Affiliations:
[a] Paediatric Unit, Department of Translational Medical Sciences, Cystic Fibrosis Regional Reference Center, University of Naples Federico II, Naples, Italy;
[b]Cystic Fibrosis Regional Reference Center, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, University of Milan, Department of Pathophysiology and Transplantation, Milan, Italy;
[c] Department of Translational Medical Science, Cystic Fibrosis Center, Adult Unit, University of Naples "Federico II", Italy;
[d]Dept of Experimental Medicine, Sapienza University of Rome, Rome, Italy;
[e]Pasteur Institute, Cenci Bolognetti Foundation, Sapienza University of Rome, Rome, Italy;
[f]Cystic Fibrosis Regional Reference Center, A.O.U. Policlinico Umberto I, Rome, Italy;
[g]Cystic Fibrosis Regional Reference Center, Mother - Child Department, United Hospitals, Ancona, Italy;
[h]Cystic Fibrosis Centre, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy;
[i] Cystic Fibrosis Regional Support Center, Department of Pediatrics, University of Brescia, ASST Spedali Civili Brescia, Brescia, Italy;
[l]Meyer Children's Hospital, Cystic Fibrosis Regional Reference Center, Department of Paediatric Medicine, Florence, Italy;
[m]Diagnostic Genetics Unit, Careggi University Hospital, Florence, Italy;
[n]IRCCS Istituto Giannina Gaslini, Genova, Italy;
[o]Cystic Fibrosis Center, Hospital San Carlo, Potenza, Italy;
[p]Freelance Statistician, Milan, Italy;

Cystic Fibrosis Research News

[q]Department of Molecular Medicine and Medical Biotechnology, University of Naples, Naples, Italy;
[r]CEINGE-Advanced Biotechnology, Naples, Italy.

What was your research question?
What is the clinical course of patients with genotype F508del/5T;TG12?

Why is this important?
5T;TG12 is a very frequent CFTR variant in some populations, especially in subjects with inconclusive diagnosis from neonatal screening (CRMS/CFSPID).

What did you do?
We have retrospectively collected clinical data of a large cohort of Italian individuals with genotype F508del/5T;TG12, followed at 10 Cystic Fibrosis Italian centers.

What did you find?
Between 129 subjects (median age: 15.0 years, range: 0–58 years; 59 older than 18 years) 30 were CF (23.3%), 41 had a mono-organ involvement (CFTR-RD)
(31.7%), and 58 had an inconclusive diagnosis after newborn screening (CRMS/CFSPID) (45.0%). After a median follow-up of 6.7 years (range 0.2-25 years), 15 patients progressed to CF, bringing the total number of CF diagnoses to 45/129 (34.9%). Most of these patients had mild lung diseases with pancreatic sufficiency and a low prevalence of CF-related complications

What does this mean and reasons for caution?
It is important to monitor the clinical picture of subjects with F508del/5T;TG12 genotype. More than one third of them develop CF during follow-up; however, they have mild lung disease and few CF-related complications.

What's next?
Continue to collect clinical data on large cohorts of subjects with little-known CFTR variant, found in individuals who are positive for neonatal screening, in order to provide useful information to families.

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