**Online Data Supplement:**
**Effects of Gender and Age of Diagnosis on Disease Progression in Long-term Cystic Fibrosis Survivors**

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**Genotype, phenotype, and demographic characteristics of the NJH population of CF patients over age 40 years**

A retrospective chart review was conducted to identify older CF patients diagnosed, evaluated or followed within the Colorado CF Center and at National Jewish Health (formerly National Jewish Medical and Research Center, Denver, Colorado). Clinical records of the Adult CF Program between 1992 and 2008 (475 patients) and the Infectious Disease Division (2822 patients) were reviewed. A cohort of 156 individuals over age 40 years at the time of last contact was analyzed. All patients included in the analysis were referred due to chronic airway infection and/or bronchiectasis, which is a characteristic clinical features of CF lung disease(1). All patients also had sweat chloride measurements diagnostic for CF (on two separate occasions) or identification of two *CFTR* mutations or variants. *CFTR* genotyping was performed in the context of clinical care over a span of nearly 20 years, thus a variety of different screens were utilized, although many patients had full sequencing of the gene. Of patients included in the Colorado database, 43.6% had two of the 23 *CFTR* mutations conclusively identified as disease causing (2), 35.9% had one disease causing mutation in association with a second variant of unknown significance, 5.1% had two mutations or variants of unknown significance, 9.6% had only a single CFTR mutation identified, and 5.8% did not undergo genotyping (**Supplemental Table E1**). For the purpose of analysis based on age of diagnosis, the childhood diagnosis (CD) was defined as patients diagnosed at age 10 or younger, while the adult diagnosis (AD) was defined as diagnosis at age 18 or older. Patients diagnosed between the ages of 10 and 18 (n=6) were not included in further analysis.

**Phenotypic consequence of 5T splicing variants in long-term CF survivors**

An abbreviated tract of five contiguous thymidines (5T) within the polythimidine locus (Tn) of intron 8 (IVS8) in *CFTR* occurs in nearly 10% of the population. When the 5T allele causes skipping of exon 9 transcription, reduced functional CFTR protein is formed (3), occasionally resulting in non-classic CF

when *in trans* with a disease causing *CFTR* mutation (4-8). The level of normal *CFTR* transcripts has been shown to correlate with the severity of pulmonary disease (9). However, penetrance of the 5T is incomplete and varies significantly between individuals (10), and even between organs of the same individual (9). In particular, the level of normal transcripts in epididymal epithelial cells is lower than the nasal epithelium, consistent with the presentation of CBAVD in the setting of subclinical lung airway disease (9). In populations of children and young adults, the presence of a 5T allele *in trans* with a known CF-causing mutation has been associated with a wide range of disease severity, but in the vast majority of cases appears to be clinically inconsequential. In the Colorado database, 20 individuals were identified with a 5T allele (**Supplemental Table E2**). From the CFF Registry, only 12 additional patients were identified, and were less completely characterized (not shown). The most common accompanying mutation was DF508 (10/20). It is known that the DF508 mutation occurs exclusively on a 9T-10TG background (11), thus patients who are carriers for DF508 and have the IVS-8 genotype 5T/9T have the 5T in *trans* with DF508 (12). In 11 of the NJH subjects, the presence of additional exonic mutations was ruled out by direct sequence analysis of all the coding exons and their corresponding splice junction sites, as well as the promoter region of the *CFTR* gene. In the remainder of the samples, the presence of common *CFTR* mutations was ruled out by various commercially available screening tests. All of these patients were diagnosed as adults (median age 54.5 yrs). Each patient had phenotypic evidence of airway disease, predominantly bronchiectasis (17/20), or sinusitis (11/20), and all underwent extensive testing to eliminate a range of other immunodeficiency disorders. In addition, all had at least one respiratory tract pathogen, including characteristic CF infections such as mucoid or nonmucoid *P. aeruginosa* (9/20) and *S. aureus* (9/20), as well as nontuberculous mycobacteria (14/20).

Four individuals in the NJH cohort were found by screening to have a 5T and 7T in combination with R117H (**Supplemental Table E2**). The age of diagnosis for these subjects ranged from 52-75 years,

and all had bronchiectasis. None of these four patients had direct sequence analysis, thus the potential exists that for any of the individuals, the R117H and 5T could be *in trans* with another (rare) *CFTR* mutation not detected by standard screening. The genotype of R117H/"Unknown" in the presence of 5T/7T has been reported in the setting of CF lung disease (13). The presence of R117H and 5T on the same allele can clearly function as a disease causing CF mutation, while R117H with 7T has variable effects when occurring *in trans* with another mutation (2, 13). In another patient, the 5T was *in trans* with a 5T allele and the R117H mutation. The genotype of 5T/5T has also been reported to result in CF lung disease in an AD patient (4).

Questions concerning the effect of gender on 5T penetrance have risen from *CFTR* screening of large populations, where penetrance of the 5T in young females has been reported to be both higher (14) and lower (11, 12) than males. In an analysis of mutation screening of 320,000 individuals, 3.8% of females compared to 42.9% of men compound heterozygous for 5T and DF508 were reported to have symptoms suggestive of non-classic CF (11, 12). The relatively low incidence of associated symptoms prompted speculation that the 5T allele has higher penetrance in males and may represent a benign variant in females (11, 12). However, an analysis of over 20,000 asymptomatic young adults presenting for pre-reproductive screening found fewer women with a 5T allele in combination with a *CFTR* mutation than predicted based on independent assembling of the two traits (14). One possible interpretation of this finding is that the penetrance of the 5T allele in combination with a *CFTR* mutation is *higher* in women, and results in a phenotype sufficiently severe to reduce the percentage of women addressing reproductive diagnostics (14).

In the 20 CF patients with a 5T over age 40 years in the NJH cohort, 80% (16/20) were women (**Supplemental Table E2**). The potential for the 5T allele to result in adult diagnosed CF was

apparent, even when the second CFTR defect was another 5T allele (n=1) or a low risk class IV or V mutation.

**References:**


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## TABLE E1: COLORADO DATABASE OF CF PATIENTS OLDER THEN 40 YEARS (1992-2008)

| Diagnosis Group[1] | Age of Diagnosis[2] | Greatest Age[2] | Sex | Outcome[3] | *CFTR* Mutations[4] | | IVS-8 | Genotype Risk Category[5] | Sweat Chloride[6] | Pancreatic Function[7] |
|---|---|---|---|---|---|---|---|---|---|---|
| CD | 0-10 | 40-50 | M | A | F508del* | F508del* | 9T/9T | HR | | PI |
| CD | 0-10 | 40-50 | F | A | G542X* | F508del* | Unknown | HR | | PI |
| CD | 0-10 | 40-50 | M | A | F508del* | F508del* | 9T/9T | HR | | PI |
| CD | 0-10 | 40-50 | M | TD | F508del* | F508del* | 9T/9T | HR | 71 | PI |
| CD | 0-10 | 40-50 | M | TD | F508del* | F508del* | 9T/9T | HR | | PI |
| CD | 0-10 | 40-50 | F | A | F508del* | F508del* | 9T/9T | HR | | PI |
| CD | 0-10 | 40-50 | M | A | F508del* | F508del* | 9T/9T | HR | | PI |
| CD | 0-10 | 40-50 | M | D | F508del* | R347P* | Unknown | LR | | PI |
| CD | 0-10 | 40-50 | M | A | F508del* | F508del* | 9T/9T | HR | | PI |
| CD | 0-10 | 40-50 | F | D | F508del* | F508del* | 9T/9T | HR | 110 | PI |
| CD | 0-10 | 40-50 | F | A | F508del* | Unknown | Unknown | Unknown | | PI |
| CD | 0-10 | 40-50 | F | A | F508del* | I507del* | Unknown | HR | | PI |
| CD | 0-10 | 40-50 | F | A | F508del* | F508del* | 9T/9T | HR | | PI |
| CD | 0-10 | 40-50 | M | D | F508del* | F508del* | 9T/9T | HR | | PI |
| CD | 0-10 | 40-50 | M | D | F508del* | Unknown | Unknown | Unknown | 106 | PI |
| CD | 0-10 | 40-50 | F | TD | Unknown | Unknown | Unknown | Unknown | 118 | PI |
| CD | 0-10 | 40-50 | M | A | F508del* | L206W | Unknown | HR | | PS |
| CD | 0-10 | 40-50 | M | A | F508del* | G551D* | 9T/9T | HR | | PI |
| CD | 0-10 | 40-50 | F | D | F508del* | R117H | Unknown | LR | | PS |
| CD | 0-10 | 40-50 | M | TD | F508del* | F508del* | 9T/9T | HR | 118 | PI |
| CD | 0-10 | 40-50 | F | D | F508del* | F508del* | 9T/9T | HR | 101 | PI |
| CD | 0-10 | 40-50 | F | A | F508del* | A455E* | 9T/9T | LR | | PS |
| CD | 0-10 | 40-50 | F | A | F508del* | F508del* | 9T/9T | HR | | PI |
| CD | 0-10 | 40-50 | M | D | F508del* | Unknown | Unknown | Unknown | 119 | PI |
| CD | 0-10 | 40-50 | F | A | F508del* | F508del* | 9T/9T | HR | 108 | PI |
| CD | 0-10 | 40-50 | F | A | W1282X* | F508del* | Unknown | HR | | PI |
| CD | 0-10 | 40-50 | M | A | F508del* | 2789+5G>A* | Unknown | LR | | PS |
| CD | 0-10 | 40-50 | F | A | F508del* | 2789+5G>A* | Unknown | LR | | PI |
| CD | 0-10 | 40-50 | M | A | F508del* | F508del* | 9T/9T | HR | | PI |
| CD | 0-10 | 40-50 | F | D | Unknown | Unknown | Unknown | Unknown | | PI |
| CD | 0-10 | 51-60 | M | A | W1282X* | F508del* | Unknown | HR | 81 | PI |
| CD | 0-10 | 51-60 | M | A | F508del* | F508del* | 9T/9T | HR | 129 | PI |
| CD | 0-10 | 51-60 | M | TD | F508del* | F508del* | 9T/9T | HR | 108 | PI |
| CD | 0-10 | 51-60 | M | A | F508del* | F508del* | 9T/9T | HR | 123 | PI |
| CD | 0-10 | 51-60 | F | D | F508del* | G551D* | 9T/9T | HR | | PI |
| CD | 0-10 | 51-60 | M | A | 621+1G>T* | F508del* | Unknown | HR | 118 | PI |
| CD | 0-10 | 51-60 | M | T | F508del* | 3849+10kbC>T* | Unknown | LR | | PS |
| CD | 0-10 | 51-60 | F | A | F508del* | S945L | Unknown | HR | | PS |

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|---|---|---|---|---|---|---|---|---|---|---|
| CD | 0-10 | 51-60 | F | D | W1282X* | 1717-1G>A* | Unknown | HR | | PI |
| CD | 0-10 | 51-60 | F | T | F508del* | F508del* | 9T/9T | HR | | PI |
| CD | 0-10 | 61-70 | M | T | 3120+1G>A* | F508del* | Unknown | HR | 68 | PS |
| N/A | 11-18 | 40-50 | F | A | F508del* | F508del* | 9T/9T | HR | | PI |
| N/A | 11-18 | 40-50 | F | A | F508del* | F508del* | 9T/9T | HR | | PI |
| N/A | 11-18 | 40-50 | M | TD | F508del* | F508del* | 9T/9T | HR | | PI |
| N/A | 11-18 | 51-60 | M | D | F508del* | F508del* | 9T/9T | HR | | PI |
| N/A | 11-18 | 51-60 | M | D | F508del* | I507del* | Unknown | HR | | PI |
| N/A | 11-18 | 61-70 | F | A | F508del* | P67L | Unknown | LR | 68 | PS |
| AD | 18-30 | 40-50 | F | A | Unknown | Unknown | Unknown | Unknown | 133 | PS |
| AD | 18-30 | 40-50 | F | A | F508del* | F508del* | 9T/9T | HR | | PI |
| AD | 18-30 | 40-50 | F | A | F508del* | 2789+5G>A* | Unknown | LR | | PS |
| AD | 18-30 | 40-50 | F | A | F508del* | D1152H | Unknown | LR | | PS |
| AD | 18-30 | 40-50 | M | D | Unknown | Unknown | Unknown | Unknown | | PI |
| AD | 18-30 | 40-50 | F | A | 1078delT | S492F | 7T/9T | LR | | PS |
| AD | 18-30 | 51-60 | F | A | F508del* | R347P* | Unknown | LR | 96 | PS |
| AD | 18-30 | 51-60 | F | TD | F508del* | 2789+5G>A* | Unknown | LR | | PS |
| AD | 31-40 | 40-50 | M | A | F508del* | L206W | Unknown | HR | 64 | PI |
| AD | 31-40 | 40-50 | F | A | W1282X* | D1152H | Unknown | LR | 33 | PS |
| AD | 31-40 | 40-50 | F | A | R1162X | Unknown | Unknown | | | PI |
| AD | 31-40 | 40-50 | F | D | F508del* | D1152H | Unknown | LR | 32 | PS |
| AD | 31-40 | 40-50 | F | A | F508del* | E384R | Unknown | LR | | PS |
| AD | 31-40 | 40-50 | F | A | F508del* | D1152H | Unknown | **LR** | | PS |
| AD | 31-40 | 40-50 | F | A | F508del* | | 5T/9T | LR | 21 | PS |
| AD | 31-40 | 40-50 | F | D | F508del* | D1152H | Unknown | LR | | PI |
| AD | 31-40 | 40-50 | F | A | F508del* | D1152H | Unknown | LR | | PS |
| AD | 31-40 | 40-50 | M | A | F508del* | | 5T/9T | LR | | PS |
| AD | 31-40 | 40-50 | M | D | F508del* | F508del* | 9T/9T | HR | | PI |
| AD | 31-40 | 40-50 | M | A | F508del* | M1101K | Unknown | LR | 115 | PI |
| AD | 31-40 | 40-50 | M | D | F508del* | F508del* | 9T/9T | HR | | PI |
| AD | 31-40 | 40-50 | M | D | F508del* | M1101K | Unknown | LR | | PI |
| AD | 31-40 | 40-50 | M | A | F508del* | R117H | Unknown | LR | | PS |
| AD | 31-40 | 40-50 | M | A | R553x | R347H | Unknown | LR | 71 | PS |
| AD | 31-40 | 40-50 | F | A | F508del* | A455E* | Unknown | LR | 94 | 0 |
| AD | 31-40 | 51-60 | M | A | F508del* | R117H | Unknown | LR | 78 | PI |
| AD | 31-40 | 61-70 | M | A | F508del* | R347H | Unknown | LR | | PS |
| AD | 41-50 | 40-50 | F | A | F508del* | D1152H | Unknown | LR | | PS |
| AD | 41-50 | 40-50 | F | A | F508del* | R117H | Unknown | LR | 39 | PS |
| AD | 41-50 | 40-50 | F | A | F508del* | 2789+5G>A* | Unknown | LR | | PS |
| AD | 41-50 | 40-50 | M | A | F508del* | | 5T/9T | LR | | PS |
| AD | 41-50 | 40-50 | F | A | W1282X* | D1152H | Unknown | LR | | PS |
| AD | 41-50 | 40-50 | F | A | S1235R | R668C | Unknown | LR | | PS |
| AD | 41-50 | 40-50 | M | A | F508del* | 2789+5G>A* | Unknown | LR | 49 | PS |
| AD | 41-50 | 40-50 | M | D | F508del* | 3849+10kbC>T* | Unknown | LR | 120 | PS |
| AD | 41-50 | 40-50 | F | TD | F508del* | 2789+5G>A* | Unknown | LR | 95 | PS |

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|---|---|---|---|---|---|---|---|---|---|---|
| AD | 41-50 | 40-50 | F | A | F508del* | R117H | Unknown | LR | 40.2 | PS |
| AD | 41-50 | 40-50 | F | A | F508del* | 3849+10kbC>T* | Unknown | LR | | PS |
| AD | 41-50 | 51-60 | F | A | F508del* | | 5T/9T | LR | | PS |
| AD | 41-50 | 51-60 | F | A | F508del* | R347H | Unknown | LR | 44 | PS |
| AD | 41-50 | 51-60 | M | A | S1235R | R668C | Unknown | LR | | PS |
| AD | 41-50 | 51-60 | F | A | F508del* | D1152H | Unknown | LR | | PS |
| AD | 41-50 | 51-60 | F | A | F508del* | R933G | 7T/9T | LR | | PS |
| AD | 41-50 | 51-60 | F | A | G542X* | A455E* | Unknown | LR | | PS |
| AD | 41-50 | 51-60 | F | A | G551D* | R117H | Unknown | LR | | PS |
| AD | 41-50 | 51-60 | M | A | Unknown | Unknown | Unknown | Unknown | 79 | PS |
| AD | 41-50 | 51-60 | F | A | F508del* | R347H | Unknown | LR | | PS |
| AD | 41-50 | 51-60 | F | A | S1251N | D1152H | Unknown | LR | | PI |
| AD | 41-50 | 51-60 | F | A | F508del* | A455E* | Unknown | LR | 97 | PS |
| AD | 41-50 | 51-60 | F | A | Unknown | Unknown | Unknown | Unknown | >60 | PI |
| AD | 41-50 | 61-70 | M | A | F508del* | R347P* | Unknown | LR | 118 | PS |
| AD | 51-60 | 51-60 | F | A | "-816C->T | | 5T/7T | LR | | PS |
| AD | 51-60 | 51-60 | F | A | F508del* | Unknown | Unknown | Unknown | 116 | PS |
| AD | 51-60 | 51-60 | M | D | R117H | | 5T/5T | LR | | PS |
| AD | 51-60 | 51-60 | F | A | 621+1G>T* | | 5T/9T | LR | | PS |
| AD | 51-60 | 51-60 | F | A | F508del* | | 5T/9T | LR | | PS |
| AD | 51-60 | 51-60 | F | A | F508del* | R117H | 7T/9T | LR | | PS |
| AD | 51-60 | 51-60 | M | A | W1282X* | D1152H | Unknown | LR | | PS |
| AD | 51-60 | 51-60 | F | A | F508del* | | **5T/9T** | LR | | PS |
| AD | 51-60 | 51-60 | M | A | W1282X* | L997F | Unknown | LR | | PS |
| AD | 51-60 | 51-60 | F | A | R117H | | 5T/7T | LR | 43 | PS |
| AD | 51-60 | 51-60 | F | A | R1162X | 1717-1G>A* | 7T/9T | LR | | PS |
| AD | 51-60 | 51-60 | F | A | F508del* | | 5T/9T | LR | | PS |
| AD | 51-60 | 51-60 | F | D | F508del* | Y109N | 7T/9T | LR | 72 | PI |
| AD | 51-60 | 51-60 | F | D | F508del* | I1027T | Unknown | LR | | PS |
| AD | 51-60 | 61-70 | M | A | F508del* | Unknown | 7T/9T | Unknown | 78 | PS |
| AD | 51-60 | 61-70 | F | A | G542X* | A455V | 7T/9T | LR | | PS |
| AD | 51-60 | 61-70 | F | A | G576A | R668C | Unknown | Unknown | | PS |
| AD | 51-60 | 61-70 | F | A | F508del* | A455E* | Unknown | LR | | PS |
| AD | 51-60 | 61-70 | F | D | F508del* | F508del* | 9T/9T | HR | 117 | PS |
| AD | 51-60 | 61-70 | M | A | F508del* | D1152H | 7T/9T | LR | | PS |
| AD | 51-60 | 61-70 | F | D | F508del* | Unknown | Unknown | Unknown | 42 | PS |
| AD | 51-60 | 61-70 | F | A | R117H | | 5T/7T | LR | | PS |
| AD | 51-60 | 61-70 | F | A | W1282X* | D1152H | Unknown | LR | | PS |
| AD | 51-60 | 61-70 | F | A | W1282X* | D1152H | Unknown | LR | | PS |
| AD | 51-60 | 61-70 | M | A | F508del* | | 5T/9T | LR | | PS |
| AD | 51-60 | 61-70 | F | D | F508del* | I148T | Unknown | LR | | PS |
| AD | 51-60 | 61-70 | F | A | W1282X* | | 5T/7T | LR | | PS |
| AD | 51-60 | 61-70 | F | A | F508del* | Unknown | Unknown | Unknown | 60.7 | PS |
| AD | 51-60 | 61-70 | M | A | F508del* | R117C | 7T/9T | LR | 96 | PS |
| AD | 51-60 | 61-70 | F | A | F508del* | 2789+5G>A* | Unknown | LR | | PS |

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|---|---|---|---|---|---|---|---|---|---|---|
| AD | 51-60 | 61-70 | F | A | q493x | l148t | Unknown | LR | 71.2 | PS |
| AD | 61-70 | 61-70 | F | A | F508del* | 406-6T>C | 7T/9T | LR | | PS |
| AD | 61-70 | 61-70 | F | A | F508del* | 1717-1G>A* | 7T/9T | LR | | PS |
| AD | 61-70 | 61-70 | M | D | F508del* | A455E* | Unknown | LR | 129 | PI |
| AD | 61-70 | 61-70 | M | A | G542X* | D1152H | 7T/9T | LR | | PS |
| AD | 61-70 | 61-70 | F | A | F508del* | I119V | Unknown | LR | | PS |
| AD | 61-70 | 61-70 | F | D | F508del* | I1027T | Unknown | LR | | PS |
| AD | 61-70 | 61-70 | F | A | F508del* | Unknown | Unknown | LR | 74.5 | PS |
| AD | 61-70 | 61-70 | F | A | G551D* | R117H | Unknown | LR | | PS |
| AD | 61-70 | 61-70 | F | A | R117H | | 5T/7T | LR | | PS |
| AD | 61-70 | 61-70 | F | A | R334W* | | 5T/7T | LR | | PS |
| AD | 61-70 | 61-70 | F | A | F508del* | C76W | 7T/9T | LR | | PS |
| AD | 61-70 | 61-70 | F | A | F508del* | R117H | Unknown | LR | | PS |
| AD | 61-70 | 61-70 | M | D | F508del* | | 5T/9T | LR | 44.7 | PS |
| AD | 61-70 | 71-80 | F | D | R560T* | D1152H | Unknown | LR | 35 | PS |
| AD | 61-70 | 71-80 | F | D | 3659delC* | Unknown | Unknown | LR | 77 | PS |
| AD | 61-70 | 71-80 | F | A | F508del* | R347H | Unknown | LR | 67 | PS |
| AD | 61-70 | 71-80 | F | A | W1282X* | | 5T/7T | LR | | PS |
| AD | 61-70 | 71-80 | F | A | F508del* | R117H | Unknown | LR | | PS |
| AD | 61-70 | 71-80 | F | A | Unknown | Unknown | Unknown | Unknown | 67 | PS |
| AD | 61-70 | 71-80 | F | A | Unknown | Unknown | Unknown | Unknown | 115 | PS |
| AD | 71-80 | 71-80 | M | D | 1898+1G>A* | R117H | Unknown | LR | | PS |
| AD | 71-80 | 71-80 | F | A | Unknown | Unknown | 7T/9T | | 61 | PS |
| AD | 71-80 | 71-80 | M | D | G542X* | R117C | Unknown | LR | 63 | PS |
| AD | 71-80 | 81-90 | F | A | R117H | | 5T/7T | LR | | PS |
| AD | 71-80 | 81-90 | F | A | F508del* | Unknown | Unknown | Unknown | 64 | PS |
| AD | 71-80 | 81-90 | M | A | F508del* | D1152H | Unknown | LR | | PS |
| AD | >80 | 81-90 | F | D | F508del* | | 5T/9T | LR | | PS |

[1]Diagnosis Group: CD= Childhood diagnosis (10 years or less), AD= Adult diagnosis (18 years or greater), N/A= Diagnosed between 10-18 years, not included in analysis.

[2]Ages are expressed as a range to protect subject anonymity.

[3]Outcome: A= Alive, D= Deceased, T= Transplanted and still living, TD= Transplanted and Deceased.

[4]*CFTR* Mutations: *mutations conclusively determined to be disease-causing.

[5]Genotype Risk Category: HR= High Risk genotype, with both CFTR mutations in Class I-III, LR= Low Risk genotype, with one or both of mutations in Class IV-V.

[6]Sweat Chloride: Mean value (mmol/L).

[7]Pancreatic Function: PI= Pancreatic Insufficient, PS= Pancreatic Sufficient.

## TABLE E2: 5T ALLELE IN LONG-TERM SURVIVORS WITH THE ADULT DIAGNOSIS OF CF (COLORADO DATABASE)

| Sex | Age[1] | Age of Dx[1] | | IV8T | *CFTR* Mutation | FEV1 (%) | Clinical features | *P. aeruginosa* | *P.A.* (mucoid) | *S. aureus* | *Stenotrophomonas maltophilia* | *Serratia marcescens* | NTM | *Aspergillus* sp. | other[2] |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| F | 40-49 | 30-39 | 5T-(TG) | 9T-(TG)10 | F508 | 47.4 | Bronchiectasis, sinusitis | | X | | | | | | |
| M | 40-49 | 30-39 | 5T | 9T | F508 | 97 | Bronchiectasis, sinusitis, DIOS[3] | | | | | | | | 6 |
| F | 40-49 | 40-49 | 5T-(TG) | 9T-(TG)10 | F508 | 108 | Sinusitis | | | X | | | X | | |
| F | 50-59 | 50-59 | 5T-(TG) | 7T-(TG)10 | *5'UTR-755* | 91 | Bronchiolitis, sinusitis | X | | | | | | | |
| F | 50-59 | 40-49 | 5T | 9T | F508 | 39 | Bronchiectasis, sinusitis, ABP/ | X | X | | X | | X | X | 7 |
| M | 50-59 | 50-59 | 5T | 5T | R117H | 22 | Bronchiectasis, sinusitis | X | X | | | | | | |
| F | 50-59 | 50-59 | 5T | 9T | *621+1G>T* | 52 | Bronchiectasis | | | X | | | X | | |
| F | 50-59 | 50-59 | 5T-(TG) | 9T-(TG)10 | F508 | 103 | Bronchiolitis, sinusitis | | | | | | X | | |
| F | 50-59 | 50-59 | 5T | 9T | F508 | 66 | Bronchiectasis, sinusitis | X | | X | | | | | |
| F | 50-59 | 50-59 | 5T | 7T | R117H | 80 | Bronchiectasis | | | | X | | X | | |
| F | 50-59 | 50-59 | 5T | 9T | F508 | 77 | Bronchiectasis | X | X | | | | X | X | |
| F | 60-69 | 50-59 | 5T | 7T | R117H | 72 | Bronchiectasis, sinusitis | X | | | | | X | | |
| M | 60-69 | 50-59 | 5T | 9T | F508 | 100 | Bronchiectasis, sinusitis, CBA | X | | X | X | X | | | 1 |
| F | 60-69 | 50-59 | 5T | 7T | *W1282X* | 58 | Bronchiectasis | | | | | X | X | | 2 |
| F | 60-69 | 60-69 | 5T | 7T | *R334W* | 109 | Bronchiectasis | | | | | | X | | |
| F | 70-79 | 60-69 | 5T | 7T | R117H | 85 | Bronchiectasis | | | | | | | | 5 |
| M | 70-79 | 60-69 | 5T | 9T | F508 | 77 | Bronchiectasis | X | | X | | | X | X | |
| F | 70-79 | 60-69 | 5T-(TG) | 7T-(TG)10 | *W1282X* | 68 | Bronchiectasis, sinusitis | | | | | | X | | 1 |
| F | 80-89 | 70-79 | 5T | 7T | R117H | 81 | Bronchiectasis | | | | | | X | | 3,4 |
| F | 80-89 | 80-89 | 5T | 9T | F508 | 82 | Bronchiectasis | | | X | | | X | | |

[1]Ages are expressed as a range to protect subject anonymity

[2]Other: (1)*Klebsiella* species, (2) *E. coli*, (3)*Pseudomonas putida*, (4)*Enterobacter* species, (5)*Citrobacter freundii*, (6)*H. influenza*, (7)*Alcaligenies xylosoxidans*

[3]DIOS: Distal Intestinal Obstruction Syndrome

[4]ABPA: Allergic bronchopulmonary aspergillosis

[5]CBAVD:Congenital bilateral absence of the vas deferens