LITERATURE REVIEW FOR ASPE B ALLELES

CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
E60X	(Strandvik, Bjorck et al. 2001)	Heterozygotes, other allele not described except in one patient, E60X/3126del4	Phenotype described for the E60X/3126del4 patient: diagnosed at 2 years, PI, moderate lung disease	Swedish	1.6
E60X	(Bienvenu, Beldjord et al. 1996)	ΔF508/E60X	Phenotype not described for this patient, but all patients in study had "classic CF."	Not described	1.7*
E60X	(Scotet, Gillet et al. 2002)	Not stated	Phenotype not described	Patients were from Brittany, France	0.67
E60X	(Sugarman, Rohlfs et al. 2004)	Not stated	Phenotypes of specific patients were not described. The characteristics of the patient population were stated as: "The CF patient population is derived from individuals referred with an indication of 'known affected' and excludes individuals referred with a 'suspected diagnosis' indication."	African American	0.46*
R75X	(Laufer-Cahana, Lerer et al. 1999)	1 patient was homozygous for R75X, the other was a compound heterozygote with the second allele not defined	Not described	Muslim Arab	3.6
R75X	(Dork, Mekus et al. 1994)	R75X/N1303K	PI	9 year old German	0.1*
R75X	(Radivojevic, Djurisic et al. 2004)	Not stated	Specific patient phenotypes were not described. "The CF diagnosis was based on typical clinical manifestations of pulmonary or/and gastrointestinal disease and high levels of sweat chloride concentration."	Patient population was from Serbia and Montenegro	0.28*

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
R75X	(Kanavakis, Efthymiadou et al. 2003)	Not stated	Not described	Greek	0.11*
406-1G>A	(Wong, Wang et al. 2001)	ΔF508/406-1G>A	Diagnosed at 4 months, severe CF, PI, poor growth, lungs colonized with *Staphylococcus*, hypersplenism, portal hypertension, liver cysts	13 year old Hispanic	1.6
		406-1G>A/unknown	Diagnosed at 7 years old, severe CF, PI, poor growth, lungs colonized with *Staphylococcus* and *Pseudomonas aeruginosa*, PPD converter	12 year old Hispanic	
406-1G>A	(Orozco, Velazquez et al. 2000)	Not stated	Specific patient phenotypes not described. All patients had CF, with diagnosis "based on abnormally elevated sweat chloride concentrations and clinical symptoms typical for CF."	Mexican	1.5
406-1G>A	(Alper, Wong et al. 2004)	7 patients were compound heterozygotes, one of which was 1288insTA/406-1G>A. The second allele was not stated for the other 6 compound heterozygotes. One patient was a homozygote.	Phenotype described for 1288insTA/406-1G>A patient: Diagnosed with CF at 4 months, FTT, persistent RTIs, clubbing, chronic congestion, cough	19 month old Mexican Hispanic female	1.1
406-1G>A	(Schrijver, Ramalingam et al. 2005)	Not stated	Phenotypes not described for this mutation. All patients in study had "clinical manifestations consistent with the spectrum of CF."	Hispanic	1.2

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
406-1G>A	(Alonso, Heine-Suner et al. 2007)	Not stated	Not described for specific patients. All patients "fulfilled the criteria of CF diagnosis."	Spanish	<0.1*
G178R	(Zielenski, Bozon et al. 1991)	G178R/ΔF508	PI	Not described	1.6*
G178R	(Cremonesi, Ferrari et al. 1992)	Not stated	Not described	Italian	0.58*
G178R	(Heim, Sugarman et al. 2001)	Not stated	Not described	7 chromosomes were from Caucasian patients, 1 chromosome was from an Asian patient	0.2% of alleles from Caucasians, 6.3% of alleles from Asians
G178R	(Sugarman, Rohlfs et al. 2004)	Not stated	Phenotypes of specific patients were not described. The characteristics of the patient population were stated as: "The CF patient population is derived from individuals referred with an indication of 'known affected' and excludes individuals referred with a 'suspected diagnosis' indication."	Hispanic	0.31*
G178R	(Castaldo, Polizzi et al. 2005)	Not stated	Specific patient phenotypes were not described. All patients in study had a diagnosis of CF "confirmed by sweat chloride levels and supported by clinical findings."	Italian	0.3*
935delA	(Wang, Bowman et al. 2000)	935delA/663delT	Diagnosed at 1 year old, severe CF with meconium ileus, PI, poor growth, early pulmonary colonization with *Pseudomonas aeruginosa*	Hispanic female, died at 4 years old	3.3
		935delA/ΔF508	Diagnosed at 2 weeks old, severe CF	8 year old Hispanic	

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
			with meconium ileus, PI, poor growth, early pulmonary colonization with *Pseudomonas aeruginosa*, GERD, liver disease, bronchopulmonary dysplasia, allergic bronchopulmonary aspergillosis	male	
935delA	(Orozco, Velazquez et al. 2000)	Not stated	Specific patient phenotypes not described. All patients had CF, with diagnosis "based on abnormally elevated sweat chloride concentrations and clinical symptoms typical for CF."	Mexican	1.0
935delA	(Wong, Wang et al. 2001)	Not stated	Severe classic clinical course, PI, poor growth	Hispanic	1.6
G330X	(Macek, Mackova et al. 1997)	Not stated	PI	African American	0.61*
G330X	(Heim, Sugarman et al. 2001)	Not stated	Not described	3 chromosomes were from African American patients, 1 was from a patient of unknown/mixed ethnicity	1.5% of African American alleles, 0.2% of unknown/mixed race alleles
G330X	(Sugarman, Rohlfs et al. 2004)	Not stated	Phenotypes of specific patients were not described. The characteristics of the patient population were stated as: "The CF patient population is derived from individuals referred with an indication of 'known affected' and excludes individuals referred with a 'suspected diagnosis' indication."	African American	0.46*
Q493X	(Kerem,	Not stated	PI	Not described	3.0*

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
	Zielenski et al. 1990)				
Q493X	(Phillips, Elias et al. 1993)	Not stated	Severe CF	Not described	Not stated
Q493X	(Kristidis, Bozon et al. 1992)	ΔF508/Q493X	PI	Not described	0.38
Q493X	(Ahmed, Corey et al. 2003)	ΔF508/Q493X	PI	Not described	0.24
Q493X	(Jones, McIntosh et al. 1992)	Not stated	Not described	Patients were of celtic and Ango-Saxon origin	4.1
1677delTA	(Koprubasi, Malik et al. 1993)	ΔF508/1677delTA	Severe CF	Turkish	1.9*
1677delTA	(Angelicheva, Boteva et al. 1994)	8 were homozygotes, 8 were compound heterozygotes with ΔF508, 2 were compound heterozygotes with an unidentified second mutation	Severe CF. 17/18 patients were diagnosed in infancy, and 9/18 died in infancy from meconium ileus or pneumonia. 17/18 were PI. 3/5 ΔF508 compound heterozygotes had liver disease. Pulmonary involvement was variable.	Patients were from the Black Sea region (Russian, Georgian, Turkish, Bulgarian, and Greek Cypriot)	1.6% of alleles overall from Black Sea region including Russia, much higher in some areas, eg. 22% of alleles from Georgia
1677delTA	(Heim, Sugarman et al. 2001)	Not stated	Not described	2 chromosomes were from Caucasian patients, 2 were from Hispanic patients	0.04% of Caucasian alleles, 0.8% of Hispanic alleles
1677delTA	(Elahi, Khodadad et al. 2006)	4 homozygotes, 1 compound heterozygotes with the second allele not described	Not described for specific mutations. Patients were diagnosed with CF based on elevated sweat chloride levels.	Iranian	7.5

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
2055del9>A	(Orozco, Zielenski et al. 1997)	2055del9>A/ΔF508, 2055del9>A/unknown	Both patients had severe CF with onset around 3 months old, PI, poor growth, moderate to severe pulmonary disease	Mexican	1.6
2055del9>A	(Alper, Wong et al. 2004)	Homozygous	Not described	Patients in study were Hispanic and African American	0.59*
2055del9>A	(Schrijver, Ramalingam et al. 2005)	Not stated	History not discussed for these patients. All patients in study had "clinical manifestations consistent with the spectrum of CF."	Hispanic	0.58
2143delT	(Dork, Kalin et al. 1992)	2143delT/ΔF508	9 months at diagnosis, PI, poor growth	10 month old German female	2.2
		2143delT/ΔF508	Diagnosed at 5 years old, PI, lungs colonized with *Pseudomonas aeruginosa*, poor growth	9 year old German female	
		2143delT/ΔF508	Diagnosed at 9 months, PI, liver disease, poor growth	16 year old German male	
		2143delT/ΔF508	Diagnosed at 2 months, PI, lungs colonized with *Pseudomonas aeruginosa*, poor growth	16 year old German male	
		2143delT/ΔF508	Diagnosed at 4 months, PI, severe liver disease, lungs colonized with *Pseudomonas aeruginosa*, poor growth	22 year old German male	
		2143delT/G551D	Diagnosed at 6 years old, PI, lungs colonized with *Pseudomonas aeruginosa*, poor growth	23 year old German male	
2143delT	(Verlingue, Kapranov et al. 1995)	Not stated	Individual patient phenotypes were not described. For all patients, diagnostic "criteria were based on two positive sweat tests and on typical findings of pulmonary disease	Russian	3.4

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
			with or without gastrointestinal disease."		
2143delT	(Heim, Sugarman et al. 2001)	Not stated	Not described	Caucasian	0.1*
K710X	(Fanen, Ghanem et al. 1992)	Not stated in the paper, but the Cystic Fibrosis Mutation Database states that the genotype of the patient was K710X/N1303K (http://www.genet.sickkids.on.ca/cftr/MutationDetailPage.external?sp=345).	The phenotype was not described in the paper, but the Cystic Fibrosis Mutation Database states that the patient was diagnosed with CF at 6 years old, and subsequently died of severe CF. The patient had pancreatic insufficiency, severe lung disease, and increased sweat chloride levels. The Cystic Fibrosis Mutation Database entry by this author also states that 4 other patients with the K710X allele and the same severe CF phenotype were identified (http://www.genet.sickkids.on.ca/cftr/MutationDetailPage.external?sp=345).	Not described.	0.48*
K710X	(Chevalier-Porst, Bonardot et al. 1994)	Not stated	Not described for individual patients. CF diagnosis was based on "2 positive sweat tests and clinical findings."	Patient population was mostly of French origin, with some mixed European and North African patients.	0.25
K710X	(Heim, Sugarman et al. 2001)	Not stated	Not described	Caucasian	0.04
K710X	(Farez-Vidal, Gomez-Llorente et al.	Not stated	Not described	Spanish	0.35*

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
	2004)				
K710X	(Claustres, Desgeorges et al. 1996)	Not stated	Not described	French	0.82
K710X	(Schibler, Bolt et al. 2001)	K710X/3905insT	Severe phenotype	Swiss	1.7*
K710X	(Tapia and Beck 2005)	ΔF508/K710X	Pancreatic insufficiency, severe failure to thrive, *Pseudomonas aeruginosa* colonization, and obstructive sleep apnea	Not described.	Case study
Q890X	(Casals, Ramos et al. 1997)	Not stated	Patient phenotypes were not described for this mutation. For the patient population as a whole, "the diagnosis was based on the clinical criteria of CF and at least two positive sweat tests."	Spanish	1.0
Q890X	(de Braekeleer, Mari et al. 1997)	ΔF508/Q890X	Diagnosed at birth, meconium ileus, bronchiectasis, allergic pulmonary aspergillosis, lungs colonized by Pseudomonas aeruginosa, PI	22 year old French Canadian male (from Saguenay Lac-St. Jean region)	Case studies
Q890X	(Sugarman, Rohlfs et al. 2004)	Not stated	Phenotypes of specific patients were not described. The characteristics of the patient population were stated as: "The CF patient population is derived from individuals referred with an indication of 'known affected' and excludes individuals referred with a 'suspected diagnosis' indication."	Hispanic	0.31*
Q890X	(Schrijver, Ramalingam et al. 2005)	Not stated	Phenotypes not described for this mutation. All patients in study had "clinical manifestations consistent	Hispanic	1.1

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
			with the spectrum of CF."		
Q890X	(Ghanem, Costes et al. 1994)	Both were Q890X/ΔF508	Both patients had classic CF, and nasal polyposis.	13 year old female, 15 year old male, both Portuguese	0.89
D1152H	(Feldmann, Rochemaure et al. 1995)	ΔF508/D1152H	Mild CF—bronchitis from childhood, moderate obstruction of lung function, no gastrointestinal symptoms	46 year old woman, ethnicity not stated	Not stated
D1152H	(Schrijver, Ramalingam et al. 2005)	Not stated	Phenotypes not described for this mutation. All patients in study had "clinical manifestations consistent with the spectrum of CF."	Hispanic	0.19*
D1152H	(Orgad, Berkenstadt et al. 2002)	D1152H/G542X	Hyperchogenic bowel, meconium ileus	Fetus at 29 weeks gestation	Case study
D1152H	(Lebecque, Leal et al. 2002)	D1152H/ΔF508	Recurrent severe pulmonary infections	7 year old Belgian male	2.3

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
		D1152H/ΔF508	Chronic cough, brochiectasis. Lobectomy, allergic bronchopulmonary aspergillosis, clubbing, bronchorrhea	18 year old Belgian female	
D1152H	(Feldmann, Couderc et al. 2003)	D1152H/R1070Q	37 years at diagnosis, CBAVD, bronchectasis	Not stated	6.7
		D1152H/ΔF508	Diagnosed at 46 years old, pulmonary symptoms		
		D1152H/ΔF508	Diagnosed at 55 years old, pulmonary symptoms, *Pseudomonas* colonization		
		D1152H/ΔF508	Diagnosed at less than 18 years old, pulmonary symptoms, PI		
D1152H	(Quint, Lerer et al. 2005)	Not stated	Individual patient phenotypes were not described, but all patients had the "classical form of CF including positive or borderline sweat test and lung disease with or without pancreatic insufficiency." The study did not include "patients (with no CF symptoms) that were referred due to congenital bilateral absence of vas deferens (CBAVD) or patients with atypical CF disease."	Ashkenazi Jewish	0.5*
D1152H	(Kornreich, Ekstein et al. 2004)	D1152H/W1282X (2 families), D1152H/ΔF508, D1152H/ 3849+10kbC>T	Phenotype described for D1152H/W1282X: digestive problems, growth retardation, no significant pulmonary problems	Ashkenazi Jewish	Frequency of allele in CF patients not stated. Carrier frequency was 12% in individuals with

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
					100% Ashkenazi Jewish descent, and 8.7% in individuals with less than 100% AJ descent
D1152H	(Highsmith, Friedman et al. 2005)	D1152H/G542X	Three siblings with mild CF. PS, mild pulmonary symptoms (cough, intermittent bronchitis), recurrent rhinosinus disease	60, 64, and 70 years old.	Case study of one family
D1152H	(Mussaffi, Prais et al. 2006)	D1152H/W1282X	Diagnosed at 46 years old. Brochiectasis, right upper lobectomy, PS, pancreatitis	54 year old Jewish male	4.9
		D1152H/D1152H	Diagnosed at 33 years old. PI, colonized with *S. aureus*	39 year old Jewish male	
		D1152H/ΔF508	Diagnosed at 41 years old. Episodes of major hemoptysis, chronic *Nocardia* infection, PS, bronchiectasis	46 year old Jewish female	
		D1152H/ΔF508	Diagnosed at 44 years old. BIPAP, bronchiectasis, PI, gallstones.	49 year old Jewish male	
		D1152H/ΔF508	Diagnosed at 49 years old. Almost no pulmonary symptoms, PI.	51 year old Jewish female	
		D1152H/D1152H	Diagnosed at 0.5 years old. PS. Some episodes of cough, abnormal chest x-rays, bacteria on sputum culture.	1.5 year old Jewish male	
		D1152H/W1282X	Diagnosed at 1.3 years old. PS. Some episodes of cough, abnormal chest x-rays, bacteria on sputum culture.	2 year old Jewish male	
		D1152H/ΔF508	Diagnosed prenatally. Persistent dilated bowel loops on prenatal ultrasound. PS, cough and rhinitis.	1 year old Jewish female	
		D1152H/ΔF508	Diagnosed prenatally. PS. Some episodes of cough, abnormal chest x-	0.8 year old Jewish male	

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
			rays, bacteria on sputum culture.		
R1158X	(Ronchetto, Telleria Orriols et al. 1992)	Second allele had an unidentified mutation	PS	Italian	0.82*
R1158X	(de Braekeleer, Mari et al. 1997)	ΔF508/R1158X	Diagnosed at birth, meconium ileus, diabetes mellitus, cholelithiasis, nasal polyps, rectal prolapse	43 year old French Canadian female (from Saguenay Lac-St. Jean region)	Case studies
R1158X	(Frossard, Abdelaziz et al. 2000)	R1158X/S549R	Mild CF. mild wheezing, intestinal problems such as diarrhea, intestinal obstruction.	16 year old male from UAE	Not applicable
R1158X	(Schrijver, Ramalingam et al. 2005)	Not stated	Phenotypes not described for this mutation. All patients in study had "clinical manifestations consistent with the spectrum of CF."	Hispanic	0.19*
R1158X	(Chillon, Casals et al. 1994)	Not stated	Not described	Spanish	0.10*
R1158X	(Claustres, Desgeorges et al. 1996)	Not stated	Not described	French	0.82
R1158X	(Castaldo, Fuccio et al. 1999)	1 homozygote, 4 heterozygotes (the second allele in the heterozygotes was not specified)	Heterozygote phenotypes not described. Homozygote was diagnosed at 3 months old and had a severe CF phenotype with FTT, severe pulmonary disease, and PI. The patient died at 20 years old.	Italian	1.3
R1158X	(Tzetis, Kanavakis et al. 1997)	Not stated	Specific patient phenotypes not described. "Diagnostic criteria involved positive sweat tests and typical clinical findings of pulmonary and gastrointestinal disease."	Greek	0.8

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
R1158X	(Kanavakis, Efthymiadou et al. 2003)	Not stated	Not described	Greek	1.0
R1158X	(Macek, Mackova et al. 1997)	Not stated	Not described	African American	0.7*
R1158X	(Heim, Sugarman et al. 2001)	Not stated	Not described	African American	2.0
R1158X	(Duarte, Amaral et al. 1996)	Complex allele R334W-R1158X, second allele was ΔF508	2 brothers were diagnosed at 3 and 8 years old, both were PS and had pulmonary problems. One brother died at 13 from cardiorespiratory insufficiency.	Portuguese	Not stated
R1158X	(Shastri, Kabra et al. 2008)	R1158X/ΔF508	Not described	Indian	0.5*
R1158X	(Sugarman, Rohlfs et al. 2004)	Not stated	Phenotypes of specific patients were not described. The characteristics of the patient population were stated as: "The CF patient population is derived from individuals referred with an indication of 'known affected' and excludes individuals referred with a 'suspected diagnosis' indication."	African American	0.46*
S1196X	(Ivaschenko, Baranov et al. 1993)	Not stated	Not described	Russian	0.63*
S1196X	(Korytina, Viktorova et al. 2002)	CFTRdele2,3/S1196R	Not described	Russian	0.8*
S1196X	(Teder,	ΔF508/S1196X	Not described	Estonian	1.7*

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
	Klaassen et al. 2000)				
S1196X	(Korytina, Viktorova et al. 2003)	S1196X/CFTRdele2,3	Not described	Russian	0.7
S1196X	(Petrova, Kapranov et al. 1997)	Two patients had the genotype S1196X/ΔF508, and the third patient had an unidentified second mutation.	Not described	Russian	1.2
S1196X	(Lakeman, Gille et al. 2008)	Not stated	Not described	Turkish	0.57*
S1196X	(Kinnunen, Bonache et al. 2005)	Not stated	Not described	Finnish	1*
CFTRdele2, 3	(Dork, Macek et al. 2000)	7 homozgotes identified	This allele was associated with severe CF. Compound heterozygotes with other severe alleles were PI and had moderate to severe lung disease. Homozygotes all had severe disease, as described below.	Seen most commonly in Central and Eastern Europeans, and sporadically in other ethnicities.	Overall frequency was not stated, but this allele is most common in Central and Eastern Europeans, with frequencies ranging from 1.1 to 6.4%.
			Diagnosed at 30 months old. PI, moderate lung disease, lungs colonized with *S. aereus*, nasal polyps.	15 year old Polish male	
			Diagnosed at 11 months. PI, moderate lung disease, lungs colonized with *P. aeruginosa*.	19 year old Polish-Canadian female	
			Diagnosed at 2 months old. Moderate lung disease, PI.	11 year old Spanish female	

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
			Diagnosed at 6 months. Meconium ileus, severe lung disease, lungs colonized with *P. aeruginosa*.	21 year old Turkish female	
			Diagnosed at birth. Meconium ileus, PI, cholestasis, extent of lung disease not documented.	7 year old German female	
			Diagnosed at 9 months. PI, nasal polyps, extent of lung disease not documented.	11 year old German male	
			Diagnosed at 4 months. PI, severe lung disease, impaired glucose tolerance test.	7 year old Czech female	
CFTRdele2, 3	(Onay, Zielenski et al. 2001)	CFTRdele2,3/ΔF508	Classical CF. PI, gastrointestinal problems, pulmonary problems, *P. aeruginosa* colonization	Turkish	0.6*
CFTRdele2, 3	(Korytina, Viktorova et al. 2002)	CFTRdele2,3/ΔF508, CFTRdele2,3/S1196X	Not described	Patients were from Bashkortostan (Russia) and were of Slavic origin	1.7
CFTRdele2, 3	(Kinnunen, Bonache et al. 2005)	Not stated	Not described	Finnish	5.9
CFTRdele2, 3	(Stanke, Ballmann et al. 2008)	Homozygous	Diagnosed at birth. Meconuim ileus, PI, lungs colonized with *P. aeruginosa*, diabetes mellitus	Not stated	Not stated
L206W	(Claustres, Desgeorges et al. 1996)	Not stated	Not described	French	5.2
L206W	(Rozen, Ferreira-Rajabi et al. 1995)	ΔF508/L206W	Asymptomatic, high sweat chloride values	30 year old French Canadian male	Case studies
		ΔF508/L206W	Sinusitis, high sweat chloride values	48 year old French Canadian female	

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
		ΔF508/L206W	Bronchiectasis, left pneumectomy, episodes of fever and cough	47 year old French Canadian female	
		Not stated for 4th adult patient	Recurrent cough and respiratory infections, *Pseudomonas aeruginusa* colonization of lungs	36 year old French Canadian female	
		Not stated for pediatric patients	Respiratory symptoms, PS	9,8, and 4 year old French Canadian children	
L206W	(Desgeorges, Rodier et al. 1995)	G542X/L206W	Diagnosed at 22 yrs, PS, normal respiratory function, hypokaliemia, diffuse muscle cramps, extracellular depletion during physical labour in hot conditions	29 year old male from Andalusia	Not stated
		ΔI507/L206W	Diagnosed at 34 yrs old, PS, had frequent upper airway infections in infancy, hypokaliemia, diffuse muscle cramps, extracellular depletion during physical labour in hot conditions	40 year old male from Southern France	
		ΔF508/L206W	Diagnosed at 15 yrs old, PS, growth retardation. Asthma, allergies, obstructive uropathy, renal cyst	17 year old female from Southern France	
		ΔF508/L206W	Diagnosed at 5 years old, chronic bronchitis, supplemented with pancreatic enzymes, mild respiratory symptoms	15 year old female from Southern France	
L206W	(des Georges, Guittard et al. 2004)	Not stated	Individual patient phenotypes were not described, but "diagnosis of classic CF [was] based on typical clinical criteria and two positive sweat tests."	French	0.7
L206W	(Heim, Sugarman et	Not stated	Not described	Caucasian	0.1*

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
	al. 2001)				
L206W	(Schrijver, Ramalingam et al. 2005)	2 patients had the genotype L206W/ΔF508, for the third patient the second allele was not described	Phenotypes not described for this mutation. All patients in study had "clinical manifestations consistent with the spectrum of CF."	Hispanic	0.58
L206W	(Bernardino, Ferri et al. 2000)	L206W/ΔF508, L206W/unknown	PS	Brazilian	0.63
L206W	(Feldmann, Couderc et al. 2003)	L206W/ΔF508, 2 patients	Diagnosed at 2 and 5 years old, normal sweat chloride, pulmonary symptoms	French	Not stated
		L206WΔ/I507	Diagnosed at 30 years old, CBAVD, normal sweat chloride, pulmonary symptoms		
L206W	(Clain, Lehmann-Che et al. 2005)	L206W/W216X	0.1 years old at diagnosis, PS, no pulmonary disease	16 year old French female	1.4
		L206W/ΔF508	0.2 years old at diagnosis, hyperechogenic fetal bowel, PS, bronchial hyperreactivity	2 year old French female	
		L206W/ΔF508	2 years old at diagnosis, PS, no pulmonary disease	16 year old French female	
		L206W/ΔF508	2 years old at diagnosis, PS, bronchitis	4 year old French male	
		L206W/ΔF508	2 years old at diagnosis, PS, bronchitis	3 year old French male	
		L206W/ΔF508	4 years old at diagnosis, PI, bronchitis	7 year old French female	
		L206W/ΔF508	5 years old at diagnosis, PS, asthma	6 year old French female	
		L206W/1342-6(T)5	28 years old at diagnosis, CBAVD, PS, bronchitis	33 year old French male	

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CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
		L206W/G542X	32 years old at diagnosis, CBAVD, PS, no pulmonary disease	43 year old French male	
		L206W/ΔF508	37 years old at diagnosis, CBAVD, other symptoms not documented	40 year old French male	
		L206W/E60X	29 years old at diagnosis, CBAVD, PS, no pulmonary disease	38 year old French male	
		L206W/ΔF508	35 years old at diagnosis, CBAVD, PI, no pulmonary disease	36 year old French male	
R1066C	(Casals, Pacheco et al. 1997)	17 patients were compound heterozygotes with ΔF508. 2 were homozygotes for R1066C. 2 were compound heterozygotes with G542X, 2 were compound heterozygotes with G542X, 2 were compound heterozygotes with 712-1G>T, 2 were compound heterozygotes with 711+1G>T, and there was 1 compound heterozygote with each of R334W and 3905insT.	All patients had a severe CF phenotype. For the compound heterozygotes, no significant differences in phenotype were found when compared to a ggroup of ΔF508 homozygotes, except a significantly higher incidence of complications such as bronchiestasis, liver disease, and nasal polyps. The two homozygotes had severe disease and died at the ages of 3 months and 7 years.	13 patients were Portuguese, 15 were Spanish	Not stated
R1066C	(Casals, Ramos et al. 1997)	Not stated	Not described	Spanish	1.1

LITERATURE REVIEW FOR ASPE B ALLELES

CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
R1066C	(Luzardo, Aznarez et al. 2002)	R1066C/ΔF508	Pulmonary symptoms	Uruguayan	0.96*
R1066C	(Liang, Wong et al. 1998)	Homozygous	Diagnosed in infancy due to FTT and recurrent pneumonia. Moderate CF, borderline PI, died at 36 from respiratory failure.	Puerto Rican.	Not stated
R1066C	(Ramirez, Ramos et al. 2006)	Not stated	Not described	Argentinian	0.97
R1066C	(Sugarman, Rohlfs et al. 2004)	Not stated	Phenotypes of specific patients were not described. The characteristics of the patient population were stated as: "The CF patient population is derived from individuals referred with an indication of 'known affected' and excludes individuals referred with a 'suspected diagnosis' indication."	Hispanic	1.9
R1066C	(Schrijver, Ramalingam et al. 2005)	Not stated	Phenotypes not described for this mutation. All patients in study had "clinical manifestations consistent with the spectrum of CF."	Hispanic	0.19*
R1066C	(Keyeux, Rodas et al. 2003)	Not stated	Individual patient phenotypes were not described. CF was diagnosed based on "clinical findings and on elevated sweat chloride concentrations."	Colombian	0.27*
W1089X	(Bernardino, Ferri et al. 2000)	R334W/W1089X	PI	Brazilian	0.31*
W1089X	(Heim, Sugarman et	Not stated	Not described	Hispanic	1.2

LITERATURE REVIEW FOR ASPE B ALLELES

CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
	al. 2001)				
W1089X	(Shoshani, Augarten et al. 1994)	One patient had the genotype W1089X/ΔF508, in the other patient the second allele was not described	PI	11 years old, Jewish	1.4
			PI, meconium ileus	7 years old, Jewish	
W1089X	(Sugarman, Rohlfs et al. 2004)	Not stated	Phenotypes of specific patients were not described. The characteristics of the patient population were stated as: "The CF patient population is derived from individuals referred with an indication of 'known affected' and excludes individuals referred with a 'suspected diagnosis' indication."	Hispanic	2.2
W1089X	(Schrijver, Ramalingam et al. 2005)	Not stated	Phenotypes not described for this mutation. All patients in study had "clinical manifestations consistent with the spectrum of CF."	Hispanic	0.39
W1089X	(Quint, Lerer et al. 2005)	Not stated	Individual patient phenotypes were not described, but all patients had the "classical form of CF including positive or borderline sweat test and lung disease with or without pancreatic insufficiency." The study did not include "patients (with no CF symptoms) that were referred due to congenital bilateral absence of vas deferens (CBAVD) or patients with atypical CF disease."	Ashkenazi Jewish	0.7
3791delC	(Macek, Mackova et	Not stated	PI	African American	0.61*

LITERATURE REVIEW FOR ASPE B ALLELES

CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
	al. 1997)				
3791delC	(Heim, Sugarman et al. 2001)	Not stated	Not described.	African American	2
3791delC	(Sugarman, Rohlfs et al. 2004)	Not stated	Not described	African American	0.46*
3199del6	(Buyse, McCarthy et al. 2004)	3199del6/G542X. This patient did not have the I148T allele, and represents the first published report of a patient carrying 3199del6 without I148T.	meconium ileus at birth, elevated sweat chloride levels, mild lung disease, pancreatic-sufficient	3 year old Hispanic American male	0.32*
3199del6	(Wong, Wang et al. 2001)	I148T/3199del6. Based on the other literature showing that I148T exists as a complex allele with 3199del6, it is likely that these two mutations were in cis in this patient, and the other chromosome contains an unidentified CF mutation.	Severe classic clinical course, PI, poor growth diabetes, lungs colonized with *E. coli* and *P. aeruginosa*	21 year old Hispanic	0.8*
3199del6	(Madore, Prevost et al. 2008)	Not stated	Not described	French Canadian	1.0
3199del6	(Claustres, Altieri et al. 2004)	3199del6/394delTT. I148T is not present.	Pancreatic insufficiency, "typical" CF lung disease, poor growth, and positive sweat test	seven years old, French	Not stated

LITERATURE REVIEW FOR ASPE B ALLELES

CFTR allele	Journal reference	Genotype of patient	CF phenotype	Patient information	Prevalence of mutation (% of alleles)
3199del6	(Rohlfs, Zhou et al. 2002)	7 patients had an allele containing I148T, 3199del6, and the 9T variant of the polythymidine tract. In trans, on the other chromosome, these patients had either ΔF508, N1303K, or Q890X.	classic CF phenotype	Not described.	Not stated.

*frequency is based on one patient or one chromosome
Definition of abbreviations: PI=pancreatic insufficient, PS=pancreatic sufficient, FTT=failure to thrive, GERD=gastrointestinal reflux disease, RTI=respiratory tract infection, CBAVD=congenital bilateral absence of the vas deferens

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