Supp. Table S1. Distribution of probes used for array-CGH analysis

Region	Chromosomal location	Size	No. of probes	Average density of probes
CFTR locus[a]	116907253-117095955[b]	189 kb	2,000	One probe every 95 bp
CFTR 5'-flanking[c]	114900000-116907253[b]	2 Mb	5,700	One probe every 350 kb
CFTR 3'-flanking[c]	117095955-117650000[b]	554 kb	2,300	One probe every 260 kb
Remaining portions of the human genome	—	3 Gb	4,000	One probe every 750 kb

[a]Defined as in (Ellsworth et al., 2000).
[b]In accordance with the human chromosome 7 assembly, March 2006 (hg18).
[c]Arbitrarily defined in this study.

Supp. Table S2. Ratio of intragenic deletion CNMs to duplication CNMs reported as a cause of inherited disease in selected human autosomal disease genes

Gene	Chromosomal location	Gene size (kb)	Disease	No. of deletions (a)	No. of duplications (b)	Ratio (a:b)
BRCA1	17q21	81	Breast and/or ovarian cancer	91	16	5.7
LDLR	19p13.2	45	Hypercholesterolemia	140	24	5.8
MLH1	3p21.3	57	Hereditary nonpolypotic colorectal cancer	76	7	10.9
MSH2	2p22-p21	80	Hereditary nonpolypotic colorectal cancer	125	5	25.0
NF1	17q11.2	283	Neurofibromatosis 1	52	4	13.0
SERPING1	11q11-q13.1	17	Angioneurotic edema	29	3	9.7
TSC2	16p13.3	41	Tuberous sclerosis	70	4	17.5

All mutations were collated from the Human Gene Mutation Database (http://www.hgmd.org).

Supp. Table S3. DNA recombination-predisposing sequence features in ±20bp flanking each of the fully characterized 40 *CFTR* intragenic breakpoints

Recombination-promoting motifs or non-B-DNA forming sequences	Actual number, in the 40 breakpoints	*P*-value
Direct repeats	7	0.001
Inverted repeats	4	0.001
Symmetric repeats	4	0.001
R-, Y- and RY-tract	446	0.0001
'Super hotspot' CCAAR	10	0.01

Refer to Table 1 and Fig. 1 for the fully characterized breakpoints.