Supplementary Data

Loss of Function HDAC8 Mutations Cause a Phenotypic Spectrum of Cornelia de Lange Syndrome-like Features, Ocular Hypertelorism, Large Fontanelle and X-linked Inheritance.

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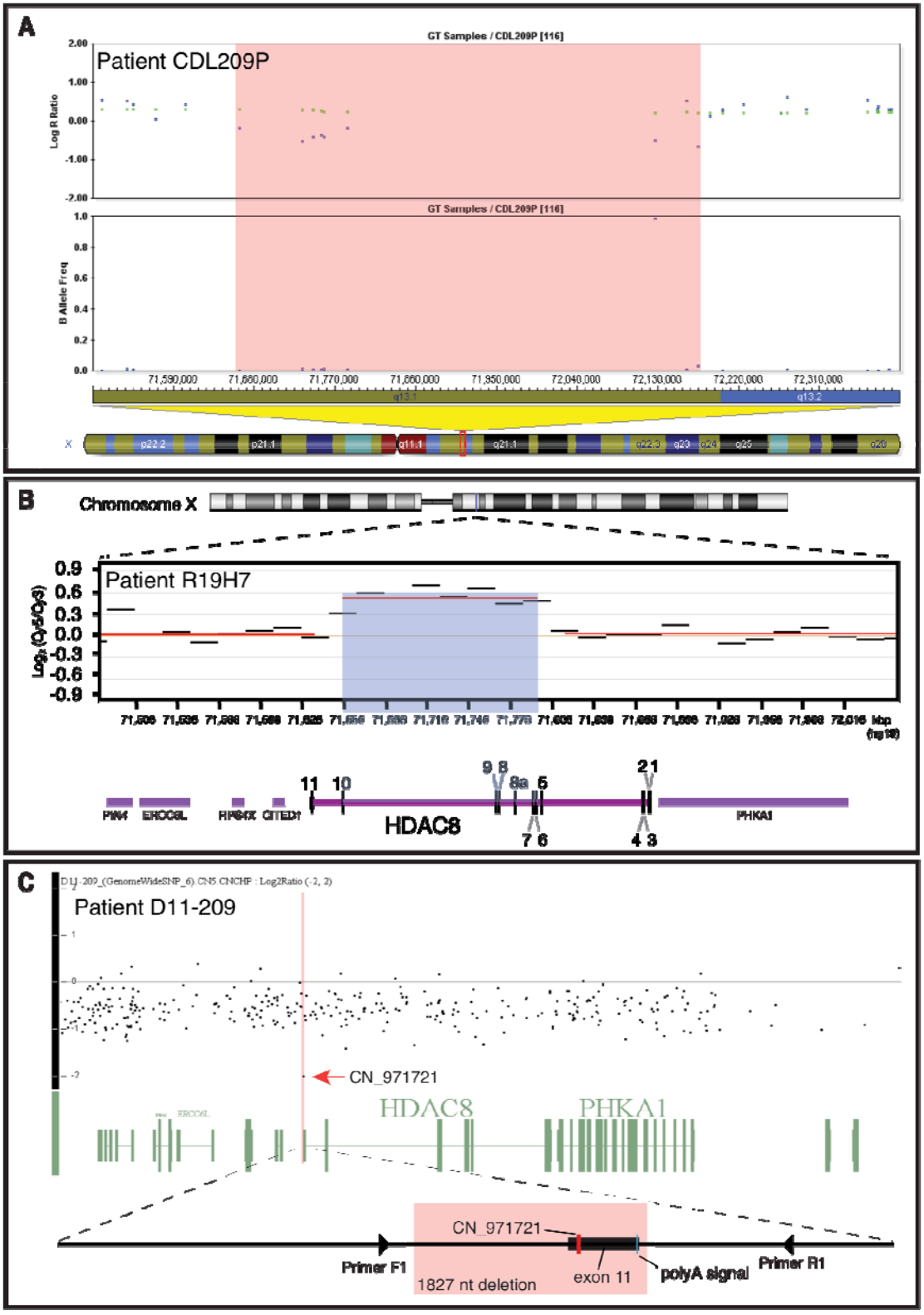

Figure S1. Chromosomal microdeletions and duplications disrupting the *HDAC8* locus. Raw data for Illumina SNP array (A), oligonucleotide array (B) and Affymetrix SNP array (C) is demonstrated. The region of chromosome or gene(s) included are shaded for the deletions (red, A,C) and duplication (blue, B).

Variant	HDAC8 activity (nmol product. μmol enzyme^{-1}.min^{-1})	Relative activity (%)
Wild-type	1390 ± 60	100 ± 4
I19S	255 ± 4	18.3 ± 0.3
H71Y	2 ± 1	0.1 ± 0.1
P91L	1220 ± 20	87.8 ± 1.4
G117E	93 ± 2	6.7 ± 0.1
C153F	34 ± 2	2.4 ± 0.1
H180R	0 ± 1	0 ± 0.1
A188T	384 ± 30	27.6 ± 2.2
D233G	416 ± 5	29.9 ± 0.4
D237Y	439 ± 6	31.6 ± 0.4
ΔK239-Y240N	0 ± 2	0 ± 0.1
I243N	475 ± 20	34.2 ± 1.4
G304R	0 ± 1	0 ± 0.1
T311M	158 ± 7	11.4 ± 0.5
G320R	234 ± 10	16.8 ± 0.7
H334R	1056 ± 40	76.0 ± 2.9

Table S1. Specific Activities and Relative Activities of HDAC8 Missense Mutations.

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