Supplemental Materials for

PhenoModifier: a genetic modifier database for elucidating the genetic basis of human phenotypic variation

Supplementary Table S1: Overview of the keywords used to search the Pubmed database.

Modify and its synonyms	Words that describe modifier effects			
modify, modifying, modified	phenotype variability, variable clinical presentation, disparate			
modifier, modifiers, modulate,	phenotypes, novel phenotype, phenotypic variation, phenotypic			
modulating, modulated,	heterogeneity, age of onset, early onset, late onset, penetrance,			
modulation	pleiotropy, expressivity, protect, protecting, protection,			
	protective, severe, severity, modest			

Screen literature	Retrieve data	⇨	Process the data
Search Pubmed using	Track down evidence		Classify modifier effects
modifier related	Strength of association		Penetrance
keywords	Odds ratio		Expressivity
Û	P value (<0.05)		Dominance
	Pedigree analysis		Pleiotropy
~10,000 papers	Gene activity study		Û
Û	Study on animal models		The modifiers and all the
	From review article		related information were
Literature clearly state	T.		loaded into MySQL.
that the studied	Modifier genes/variants		Û
gene(s)/variant(s)	Target disorders		Design the
function as modifier	Modifier effects	P	PhenoModifier database

Supplementary Figure S1: Schematic representation of the data curation and processing procedure.