

**TABLE 26.1.** Relative frequency of types of mutations underlying disease phenotypes

Change	Number	Percent of Total
Deletion	6,085	21.8
Insertion/duplication	1,911	6.8
Complex rearrangement	512	1.8
Repeat variations	38	0.1
Missense/nonsense	16,441	58.9
Splicing	2,727	9.8
Regulatory	213	0.8
Total	27,027	100.0

\*Data are from the Human Gene Mutation Database (June 2002); <http://www.hgmd.cf.ac.uk>.