Human genes associated with disease / syndromes by missense mutations:

Phenylketonuria: Phenylalanine hydroxylase (PAH)
I65T
R158Q
R261Q
P281L
R408W

BH4-deficient hyperphenylalaninemia (HPA) D [HPABH4D]: PCBD1 (pterin carbinolamine dehydratase)
T79I
C83R
R88Q

Tay Sachs disease: Beta-hexosaminidase A (HEXA)
P25S
L39R
L127R
R170W
R178C
R178H
K197T
H204R

Sandhoff disease: Beta-hexosaminidase B (HEXB)
W57C
T150P
Y266D
R505Q

Ehlers-Danlos syndrome with short stature and limb anomalies (EDSSLA) - 4-Beta-Galactosyltransferase (B4GALT7)
L41P
R141W
A186D
L206P
C214Y
R270C

Gaucher disease (various types): Acid beta-glucocerebrosidase aka beta-glucosidase (GBA)
N370S
V394L
P415R
L444P
L483P

Severe combined immunodeficiency: Adenosine deaminase (ADA)

R156C
R156H
G216R
S291L
A329V

Atelosteogenesis (various types), Boomerang Dysplasia: filamin B (FLNB)
L171R
A173T
M202V
S235P
G751R

Tangier disease: ATP-binding cassette Transporter1 (ABCA1)
W530S
N875S
A877V
D1099Y
S1446L

Hutchinson-Gilford progeria syndrome: Lamin A (LMNA)
R133L
E145K
R471C
R527C
G608S