

Comparison of Clinical Features of Timothy Syndrome Variants

Clinical Findings	TS1 G406R (exon 8A)	TS2 G402S and G406R (exon 8)	TS3 A1473G
QT prolongation	Yes	Yes	Yes
Syndactyly of hands or feet	Yes	No	Yes
	Yes	No	Yes
Congenital cardiac defects	Yes	No	Yes
Cardiac hypertrophy	Yes	Yes	No
Autism, mental retardation, delayed development	Yes	?	Yes
Round face	Yes	Yes	Yes
Sparse hair	Yes	Yes	Yes
Sparse teeth	Yes	Yes	Yes
Low-set ears	Yes	Yes	Yes
Flat nasal bridge	Yes	Yes	Yes
Mandibular hypoplasia	Yes	Yes	Yes
Downturned lips corner, thin upper lips	Yes	Yes	Yes
Stroke	No	No	Yes
Cortical blindness	No	No	Yes
Nemaline myopathy	No	Yes*	Yes
Joint contracture	No	No	Yes
Dislocated hips	No	Yes	Yes
Hypoglycemia	Yes	No	Yes
Hypocalcemia	Yes	No	Yes