

ENDOCRINE TUMOR

GENE PANEL DG 3.6.0 (7 GENES)

Releasedate: 05-04-2023

<i>Gene</i>	<i>TWIST X2 covered >10x</i>	<i>TWIST X2 covered >20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
AIP	100%	100%	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200
CDC73	100%	100%	Hyperparathyroidism, familial primary, 145000 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266 Hyperparathyroidism-jaw tumor syndrome, 145001
CDKN1A	100%	100%	No OMIM disease ID
CDKN1B	100%	100%	Multiple endocrine neoplasia, type IV, 610755
CDKN2B	100%	100%	No OMIM disease ID
CDKN2C	100%	100%	No OMIM disease ID
MEN1	100%	100%	Multiple endocrine neoplasia 1, 131100 Lipoma, somatic, Angiofibroma, somatic, Carcinoid tumor of lung, Adrenal adenoma, somatic, Parathyroid adenoma, somatic,

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

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TWIST X2 is the chemistry used for WES analysis.

Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with coverage denoting NC are non-protein coding genes.

non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : March 17th, 2023

This list is accurate for panel version DG 3.6.0

Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors
