

Supplementary information S1 | **Mutations of miRNA biogenesis pathway components in human cancers**

Location	Mutation	Number	Type	Cancer Type
DROSHA				
Q46*	Non-sense	3	Somatic	Wilms tumors ^{1, 2}
M120V	Mis-sense	1	Gemline	Wilms tumors ³
P211T	Mis-sense	1	Somatic	Wilms tumors ¹
R279C	Mis-sense	2	Gemline	Wilms tumors ⁴
R414*	Non-sense	3	Somatic	Wilms tumors ^{1, 2}
E696V	Mis-sense	2	Somatic	Wilms tumors ²
L728V	Mis-sense	1	Somatic	Wilms tumors ³
R967W	Mis-sense	1	Gemline	Wilms tumors ³
E969K	Mis-sense	2	Somatic	Wilms tumors ²
D973H	Mis-sense	1	Somatic	Wilms tumors ⁴
S990R	Mis-sense	1	Somatic	Wilms tumors ⁴
E993K	Mis-sense	5	Somatic	Wilms tumors ^{2, 4}
E1147K	Mis-sense	81	Somatic	Wilms tumors ¹⁻⁴
E1147V	Mis-sense	1	Somatic	Wilms tumors ²
D1151Y	Mis-sense	1	Somatic	Wilms tumors ³
D1151A	Mis-sense	2	Somatic	Wilms tumors ²
D1151H	Mis-sense	1	Somatic	Wilms tumors ²
D1151G	Mis-sense	5	Somatic	Wilms tumors ^{1, 2}
Q1186K	Mis-sense	1	Somatic	Wilms tumors ⁴
Q1187K	Mis-sense	5	Somatic	Wilms tumors ²
D1204Y	Mis-sense	1	Somatic	Wilms tumors ⁴
E1222K	Mis-sense	2	Somatic	Wilms tumors ^{2, 4}
E1222G	Mis-sense	2	Somatic	Wilms tumors ²
I1225M	Mis-sense	1	Somatic	Wilms tumors ⁴
DGCR8				
R32fs	Frame-shift	1	Somatic	Wilms tumors ¹
G55S	Mis-sense	1	Somatic	Wilms tumors ¹
G71*	Non-sense	1	Somatic	Wilms tumors ²
K82*	Non-sense	1	Somatic	Wilms tumors ⁴
S92R	Mis-sense	1	Somatic	Wilms tumors ¹

E213*	Non-sense	1	Somatic	Wilms tumors ⁴
Y239*	Non-sense	1	Somatic	Wilms tumors ²
E518K	Mis-sense	45	Somatic	Wilms tumors ^{1, 2, 4}
A558T	Mis-sense	1	Somatic	Wilms tumors ¹
K588R	Mis-sense	2	Germline	Wilms tumors ⁴
L694S	Mis-sense	1	Somatic	Wilms tumors ²
S720N	Mis-sense	1	Germline	Wilms tumors ³
Y721H	Mis-sense	1	Somatic	Wilms tumors ¹
XPO5				
C50R	Mis-sense	1	Somatic	Wilms tumors ²
R159*	Non-sense	2	Germline	Wilms tumors ²
Y316*	Non-sense	1	Somatic	Wilms tumors ²
R440*	Non-sense	2	Somatic	Wilms tumors ²
R440Q	Mis-sense	1	Somatic	Wilms tumors ²
Q572fs	Frame-shift	1	Somatic	Wilms tumors ²
E822A	Mis-sense	1	Somatic	Wilms tumors ²
V832I	Mis-sense	1	Somatic	Wilms tumors ¹
L843P	Mis-sense	1	Somatic	Wilms tumors ²
L927P	Mis-sense	1	Somatic	Wilms tumors ²
S1125*	Non-sense	1	Somatic	Wilms tumors ²
R1167fs	Frame-shift	8	Non-determined	Mix tumors ⁵
F1179fs	Frame-shift	30	Non-determined	Mix tumors ⁵
K1181fs	Frame-shift	39	Non-determined	Mix tumors ⁵
TARBP2				
M145fs	Frame-shift	9	Non-determined	Mix tumors ⁶
P151fs	Frame-shift	63	Non-determined	Mix tumors ⁶
D221G	Mis-sense	1	Somatic	Wilms tumors ³
R296H	Mis-sense	1	Somatic	Wilms tumors ¹
R353fs	Frame-shift	1	Somatic	Wilms tumors ¹
DICER1				
R187*	Non-sense	1	Germline	Pleuropulmonary blastoma ⁷
I383fs	Frame-shift	1	Non-determined	Pleuropulmonary blastoma ⁸
S436fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁹
R459fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁷
I461fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁸

E493*	Non-sense	1	Gemline	Pleuropulmonary blastoma ¹⁰
R534*	Non-sense	1	Gemline	Pleuropulmonary blastoma ¹⁰
R544*	Non-sense	1	Germline	Pleuropulmonary blastoma ⁷
M552fs	Frame-shift	1	Gemline	Pleuropulmonary blastoma ¹⁰
M562fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁷
Y627*	Non-sense	1	Gemline	Pleuropulmonary blastoma ¹⁰
R646*	Non-sense	1	Gemline	Pleuropulmonary blastoma ¹⁰
R656*	Non-sense	1	Germline	Pleuropulmonary blastoma ⁷
V680fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁷
Y739*	Non-sense	1	Gemline	Pleuropulmonary blastoma ¹⁰
P740fs	Frame-shift	1	Gemline	Pleuropulmonary blastoma ¹⁰
T788fs	Frame-shift	1	Gemline	Pleuropulmonary blastoma ¹⁰
R934*	Non-sense	1	Gemline	Pleuropulmonary blastoma ¹⁰
T955fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁸
R1003*	Non-sense	1	Non-determined	Pleuropulmonary blastoma ⁸
Y1091*	Non-sense	1	Germline	Pleuropulmonary blastoma ⁷
P1161fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁸
Y1170*	Non-sense	1	Gemline	Pleuropulmonary blastoma ¹⁰
C1197fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁷
S1250fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁸
L1469fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁷
W1506*	Non-sense	1	Germline	Pleuropulmonary blastoma ⁷
E1519*	Non-sense	1	Germline	Pleuropulmonary blastoma ¹¹
L1573R	Mis-sense	1	Gemline	Pleuropulmonary blastoma ¹⁰
S1637*	Non-sense	1	Non-determined	Pleuropulmonary blastoma ⁸
D1654fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁷
E1705K	Mis-sense	8	Somatic	Pleuropulmonary blastoma ⁷
E1705V	Mis-sense	1	Non-determined	Pleuropulmonary blastoma ⁸
G1708E	Mis-sense	1	Somatic	Pleuropulmonary blastoma ⁷
D1709N	Mis-sense	7	Somatic	Pleuropulmonary blastoma ^{7, 8}
K1798fs	Frame-shift	1	Germline	Pleuropulmonary blastoma ⁷
G1809R	Mis-sense	24	Somatic	Pleuropulmonary blastoma ^{7, 8}
D1810Y	Mis-sense	5	Somatic	Pleuropulmonary blastoma ^{7, 8}
E1813D	Mis-sense	2	Somatic	Pleuropulmonary blastoma ⁷
E1813G	Mis-sense	7	Somatic	Pleuropulmonary blastoma ^{7, 8, 11}

E1813K	Mis-sense	1	Somatic	Pleuropulmonary blastoma ⁷
E1813Q	Mis-sense	1	Somatic	Pleuropulmonary blastoma ⁷
Y1820*	Non-sense	1	Germline	Pleuropulmonary blastoma ⁸
E503*	Non-sense	1	Germline	Nonepithelial ovarian cancers ¹²
R459fs	Frame-shift	1	Germline	Nonepithelial ovarian cancers ¹²
C748fs	Frame-shift	1	Germline	Nonepithelial ovarian cancers ¹²
Y819*	Non-sense	1	Germline	Nonepithelial ovarian cancers ¹³
P942*	Non-sense	1	Germline	Nonepithelial ovarian cancers ¹³
E1025*	Non-sense	2	Germline	Nonepithelial ovarian cancers ¹²
G1079*	Non-sense	1	Somatic	Nonepithelial ovarian cancers ¹³
Y1204*	Non-sense	1	Germline	Nonepithelial ovarian cancers ¹³
V1260fs	Frame-shift	1	Germline	Nonepithelial ovarian cancers ¹²
D1699fs	Frame-shift	1	Germline	Nonepithelial ovarian cancers ¹²
R1703*	Non-sense	1	Germline	Nonepithelial ovarian cancers ¹³
E1705K	Mis-sense	6	Somatic	Nonepithelial ovarian cancers ^{13, 14}
D1709N	Mis-sense	13	Somatic	Nonepithelial ovarian cancers ^{13, 14}
D1709N	Mis-sense	1	Non-determined	Nonepithelial ovarian cancers ¹²
D1709E	Mis-sense	3	Somatic	Nonepithelial ovarian cancers ¹³
D1709G	Mis-sense	3	Somatic	Nonepithelial ovarian cancers ¹³
E1788fs	Frame-shift	5	Somatic	Nonepithelial ovarian cancers ¹⁴
D1810H	Mis-sense	1	Somatic	Nonepithelial ovarian cancers ¹³
D1810N	Mis-sense	1	Somatic	Nonepithelial ovarian cancers ¹³
D1810Y	Mis-sense	4	Somatic	Nonepithelial ovarian cancers ^{13, 14}
D1810V	Mis-sense	1	Somatic	Nonepithelial ovarian cancers ¹⁴
E1813A	Mis-sense	1	Non-determined	Nonepithelial ovarian cancers ¹²
E1813D	Mis-sense	2	Somatic	Nonepithelial ovarian cancers ¹⁴
E1813K	Mis-sense	2	Somatic	Nonepithelial ovarian cancers ^{13, 14}
E1813G	Mis-sense	2	Somatic	Nonepithelial ovarian cancers ¹³
E1813Q	Mis-sense	4	Somatic	Nonepithelial ovarian cancers ^{13, 14}
W1831*	Non-sense	1	Somatic	Nonepithelial ovarian cancers ¹³
Q48E	Mis-sense	1	Somatic	Wilms tumour ¹
I85M	Mis-sense	1	Somatic	Wilms tumour ¹
I85M	Mis-sense	1	Germline	Wilms tumors ²
R307fs	Frame-shift	4	Germline	Wilms tumors ^{9, 15}
S436fs	Frame-shift	2	Germline	Wilms tumors ^{9, 15}

P645*	Non-sense	2	Somatic	Wilms tumors ¹⁵
G706fs	Frame-shift	4	Germline	Wilms tumors ^{9, 15}
A872T	Mis-sense	1	Germline	Wilms tumors ¹⁵
L999*	Non-sense	3	Somatic	Wilms tumors ¹⁵
R1003Q	Mis-sense	1	Germline	Wilms tumors ²
A1011*	Non-sense	1	Somatic	Wilms tumors ¹⁵
R1071*	Non-sense	1	Somatic	Wilms tumors ¹⁵
I1102fs	Frame-shift	1	Somatic	Wilms tumors ³
I1110V	Mis-sense	1	Non-determined	Wilms tumors ¹⁵
K1324*	Non-sense	2	Somatic	Wilms tumors ¹⁵
S1344L	Mis-sense	1	Somatic	Wilms tumors ¹⁵
R1386C	Mis-sense	1	Germline	Wilms tumors ²
S1505*	Non-sense	1	Somatic	Wilms tumors ¹⁵
A1560*	Non-sense	2	Somatic	Wilms tumors ¹⁵
H1693N	Mis-sense	1	Germline	Wilms tumors ²
E1705K	Mis-sense	2	Germline	Wilms tumors ²
D1709N	Mis-sense	2	Germline	Wilms tumors ²
D1713A	Mis-sense	1	Somatic	Wilms tumors ¹⁵
D1713V	Mis-sense	1	Germline	Wilms tumors ²
L1777H	Mis-sense	1	Germline	Wilms tumors ¹⁵
E1788fs	Frame-shift	2	Somatic	Wilms tumors ¹⁵
G1809R	Mis-sense	1	Somatic	Wilms tumors ³
G1809V	Mis-sense	1	Somatic	Wilms tumors ³
D1810N	Mis-sense	1	Somatic	Wilms tumour ¹
D1810N	Mis-sense	2	Germline	Wilms tumors ²
A1818T	Mis-sense	1	Somatic	Wilms tumors ¹⁵
G1886R	Mis-sense	1	Germline	Wilms tumors ⁴
K429fs	Frame-shift	1	Germline	Pituitary blastoma ¹⁶
R509*	Non-sense	1	Germline	Pituitary blastoma ¹⁶
R676*	Non-sense	2	Germline	Pituitary blastoma ¹⁶
Y793*	Non-sense	1	Germline	Pituitary blastoma ¹⁶
N1093*	Non-sense	1	Germline	Pituitary blastoma ¹⁶
S1179fs	Frame-shift	1	Germline	Pituitary blastoma ¹⁶
D1437fs	Frame-shift	1	Germline	Pituitary blastoma ¹⁶
D1709H	Mis-sense	1	Germline	Pituitary blastoma ¹⁶

D1709T	Mis-sense	1	Somatic	Pituitary blastoma ¹⁶
D1709N	Mis-sense	1	Somatic	Pituitary blastoma ¹⁶
G1809W	Mis-sense	1	Somatic	Pituitary blastoma ¹⁶
E1813D	Mis-sense	1	Somatic	Pituitary blastoma ¹⁶
E1813V	Mis-sense	1	Somatic	Pituitary blastoma ¹⁶
E1813K	Mis-sense	2	Somatic	Pituitary blastoma ¹⁶
Y793*	Non-sense	1	Germline	Differentiated thyroid arcinoma ¹⁷
S1169fs	Frame-shift	1	Germline	Differentiated thyroid carcinoma ¹⁷
N1193fs	Frame-shift	1	Germline	Differentiated thyroid carcinoma ¹⁷
E1705K	Mis-sense	1	Somatic	Differentiated thyroid carcinoma ¹⁷
E1813D	Mis-sense	1	Somatic	Differentiated thyroid carcinoma ¹⁷
E1813G	Mis-sense	1	Somatic	Differentiated thyroid carcinoma ¹⁷
Q249fs	Frame-shift	1	Non-determined	Childhood cystic nephroma ¹⁸
S396R	Mis-sense	1	Non-determined	Childhood cystic nephroma ¹⁸
K429fs	Frame-shift	1	Non-determined	Childhood cystic nephroma ¹⁸
R509*	Non-sense	1	Non-determined	Childhood cystic nephroma ¹⁸
P817fs	Frame-shift	1	Non-determined	Childhood cystic nephroma ¹⁸
Y976*	Non-sense	1	Non-determined	Childhood cystic nephroma ¹⁸
V990fs	Frame-shift	1	Non-determined	Childhood cystic nephroma ¹⁸
L1012fs	Frame-shift	1	Non-determined	Childhood cystic nephroma ¹⁸
Q1031*	Non-sense	1	Non-determined	Childhood cystic nephroma ¹⁸
K1324fs	Frame-shift	1	Non-determined	Childhood cystic nephroma ¹⁸
Y1335*	Non-sense	1	Non-determined	Childhood cystic nephroma ¹⁸
P1336fs	Frame-shift	1	Non-determined	Childhood cystic nephroma ¹⁸
D1437fs	Frame-shift	4	Germline	Familial cystic nephroma ¹⁹
E1705K	Mis-sense	3	Non-determined	Childhood cystic nephroma ¹⁸
D1709N	Mis-sense	3	Non-determined	Childhood cystic nephroma ¹⁸
E1778fs	Frame-shift	1	Non-determined	Childhood cystic nephroma ¹⁸
G1809R	Mis-sense	2	Non-determined	Childhood cystic nephroma ¹⁸
D1810H	Mis-sense	1	Non-determined	Childhood cystic nephroma ¹⁸
E1813D	Mis-sense	5	Non-determined	Childhood cystic nephroma ¹⁸
E1813G	Mis-sense	3	Non-determined	Childhood cystic nephroma ¹⁸
E1813K	Mis-sense		Non-determined	Childhood cystic nephroma ¹⁸
S1826*	Non-sense	5	Germline	Familial cystic nephroma ¹⁹
Y637*	Non-sense	1	Germline	Embryonal rhabdomyosarcoma (ERMS) ²⁰

G706fs	Frame-shift	3	Germline	Cervix embryonal rhabdomyosarcoma ⁹
Y749*	Non-sense	1	Germline	Embryonal rhabdomyosarcoma (ERMS) ²⁰
S1179fs	Frame-shift	1	Germline	Embryonal Rhabdomyosarcoma ²¹
Y1204fs	Frame-shift	3	Germline	Cervix embryonal rhabdomyosarcoma ⁹
L1303*	Non-sense	1	Germline	Rhabdomyosarcoma ¹³
L1303fs	Frame-shift	4	Germline	Cervix embryonal rhabdomyosarcoma ⁹
E1418_E1420del	Deletion	1	Somatic	Embryonal rhabdomyosarcoma (ERMS) ²⁰
D1437fs	Frame-shift	1	Germline	Embryonal rhabdomyosarcoma (ERMS) ²⁰
Q1702*	Non-sense	1	Germline	Embryonal rhabdomyosarcoma (ERMS) ²⁰
E1705K	Mis-sense	1	Somatic	Rhabdomyosarcoma ¹³
L1789fs	Frame-shift	1	Somatic	Embryonal rhabdomyosarcoma (ERMS) ²⁰
E1813K	Mis-sense	1	Somatic	Embryonal Rhabdomyosarcoma ²¹
P291*	Non-sense	4	Germline	Nontoxic multinodular goiter ²²
R509*	Non-sense	1	Non-determined	Multinodular goiter ²³
S839F	Mis-sense	20	Germline	Nontoxic multinodular goiter ²²
I813_Y819del	Deletion	3	Germline	Nontoxic multinodular goiter ²²
R935_R996 del	Deletion	8	Germline	Nontoxic multinodular goiter ²²
R1672*	Non-sense	2	Germline	Nontoxic multinodular goiter ²²
N1742_F1745del	Deletion	2	Germline	Multinodular Goiter ²⁴
R656*	Non-sense	4	Germline	Pulmonary sequestration ⁹
V1351fs	Frame-shift	1	Germline	Primitive neuroectodermal tumor ⁹
F1706L	Mis-sense	1	Non-determined	Non-small cell lung cancers ²⁵
L1712fs	Frame-shift	1	Non-determined	Gastric carcinoma ²⁵
E1801fs	Frame-shift	1	Non-determined	Prostate carcinoma ²⁵
E1797D	Mis-sense	1	Non-determined	Soft tissue sarcoma ²⁵

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