*SOLUCIONS*

GENE IDH2

***IDH2 (ENST00000330062.8)***

***WILD TYPE DNA IDH2***

**Nucleotide Sequence (1359 nt):**

>IDH2
ATGGCCGGCTACCTGCGGGTCGTGCGCTCGCTCTGCAGAGCCTCAGGCTCGCGGCCGGCCTGGGCGCCGG
CGGCCCTGACAGCCCCCACCTCGCAAGAGCAGCCGCGGCGCCACTATGCCGACAAAAGGATCAAGGTGGC
GAAGCCCGTGGTGGAGATGGATGGTGATGAGATGACCCGTATTATCTGGCAGTTCATCAAGGAGAAGCTC
ATCCTGCCCCACGTGGACATCCAGCTAAAGTATTTTGACCTCGGGCTCCCAAACCGTGACCAGACTGATG
ACCAGGTCACCATTGACTCTGCACTGGCCACCCAGAAGTACAGTGTGGCTGTCAAGTGTGCCACCATCAC
CCCTGATGAGGCCCGTGTGGAAGAGTTCAAGCTGAAGAAGATGTGGAAAAGTCCCAATGGAACTATCCGG
AACATCCTGGGGGGGACTGTCTTCCGGGAGCCCATCATCTGCAAAAACATCCCACGCCTAGTCCCTGGCT
GGACCAAGCCCATCACCATTGGCAGGCACGCCCATGGCGACCAGTACAAGGCCACAGACTTTGTGGCAGA
CCGGGCCGGCACTTTCAAAATGGTCTTCACCCCAAAAGATGGCAGTGGTGTCAAGGAGTGGGAAGTGTAC
AACTTCCCCGCAGGCGGCGTGGGCATGGGCATGTACAACACCGACGAGTCCATCTCAGGTTTTGCGCACA
GCTGCTTCCAGTATGCCATCCAGAAGAAATGGCCGCTGTACATGAGCACCAAGAACACCATACTGAAAGC
CTACGATGGGCGTTTCAAGGACATCTTCCAGGAGATCTTTGACAAGCACTATAAGACCGACTTCGACAAG
AATAAGATCTGGTATGAGCACCGGCTCATTGATGACATGGTGGCTCAGGTCCTCAAGTCTTCGGGTGGCT
TTGTGTGGGCCTGCAAGAACTATGACGGAGATGTGCAGTCAGACATCCTGGCCCAGGGCTTTGGCTCCCT
TGGCCTGATGACGTCCGTCCTGGTCTGCCCTGATGGGAAGACGATTGAGGCTGAGGCCGCTCATGGGACC
GTCACCCGCCACTATCGGGAGCACCAGAAGGGCCGGCCCACCAGCACCAACCCCATCGCCAGCATCTTTG
CCTGGACACGTGGCCTGGAGCACCGGGGGAAGCTGGATGGGAACCAAGACCTCATCAGGTTTGCCCAGAT
GCTGGAGAAGGTGTGCGTGGAGACGGTGGAGAGTGGAGCCATGACCAAGGACCTGGCGGGCTGCATTCAC
GGCCTCAGCAATGTGAAGCTGAACGAGCACTTCCTGAACACCACGGACTTCCTCGACACCATCAAGAGCA
ACCTGGACAGAGCCCTGGGCAGGCAGTAG

**Translation (452 aa):**

>IDH2
MAGYLRVVRSLCRASGSRPAWAPAALTAPTSQEQPRRHYADKRIKVAKPVVEMDGDEMTRIIWQFIKEKL
ILPHVDIQLKYFDLGLPNRDQTDDQVTIDSALATQKYSVAVKCATITPDEARVEEFKLKKMWKSPNGTIR
NILGGTVFREPIICKNIPRLVPGWTKPITIGRHAHGDQYKATDFVADRAGTFKMVFTPKDGSGVKEWEVY
NFPAGGVGMGMYNTDESISGFAHSCFQYAIQKKWPLYMSTKNTILKAYDGRFKDIFQEIFDKHYKTDFDK
NKIWYEHRLIDDMVAQVLKSSGGFVWACKNYDGDVQSDILAQGFGSLGLMTSVLVCPDGKTIEAEAAHGT
VTRHYREHQKGRPTSTNPIASIFAWTRGLEHRGKLDGNQDLIRFAQMLEKVCVETVESGAMTKDLAGCIH
GLSNVKLNEHFLNTTDFLDTIKSNLDRALGRQ

***TUMOR DNA (Bone Marrow)***

**Nucleotide Sequence (1359 nt):**

>IDH2
ATGGCCGGCTACCTGCGGGTCGTGCGCTCGCTCTGCAGAGCCTCAGGCTCGCGGCCGGCCTGGGCGCCGG
CGGCCCTGACAGCCCCCACCTCGCAAGAGCAGCCGCGGCGCCACTATGCCGACAAAAGGATCAAGGTGGC
GAAGCCCGTGGTGGAGATGGATGGTGATGAGATGACCCGTATTATCTGGCAGTTCATCAAGGAGAAGCTC
ATCCTGCCCCACGTGGACATCCAGCTAAAGTATTTTGACCTCGGGCTCCCAAACCGTGACCAGACTGATG
ACCAGGTCACCATTGACTCTGCACTGGCCACCCAGAAGTACAGTGTGGCTGTCAAGTGTGCCACCATCAC
CCCTGATGAGGCCCGTGTGGAAGAGTTCAAGCTGAAGAAGATGTGGAAAAGTCCCAATGGAACTATCCGG
AACATCCTGGGGGGGACTGTCTTCCGGGAGCCCATCATCTGCAAAAACATCCCACGCCTAGTCCCTGGCT
GGACCAAGCCCATCACCATTGGC**AAG**CACGCCCATGGCGACCAGTACAAGGCCACAGACTTTGTGGCAGA
CCGGGCCGGCACTTTCAAAATGGTCTTCACCCCAAAAGATGGCAGTGGTGTCAAGGAGTGGGAAGTGTAC
AACTTCCCCGCAGGCGGCGTGGGCATGGGCATGTACAACACCGACGAGTCCATCTCAGGTTTTGCGCACA
GCTGCTTCCAGTATGCCATCCAGAAGAAATGGCCGCTGTACATGAGCACCAAGAACACCATACTGAAAGC
CTACGATGGGCGTTTCAAGGACATCTTCCAGGAGATCTTTGACAAGCACTATAAGACCGACTTCGACAAG
AATAAGATCTGGTATGAGCACCGGCTCATTGATGACATGGTGGCTCAGGTCCTCAAGTCTTCGGGTGGCT
TTGTGTGGGCCTGCAAGAACTATGACGGAGATGTGCAGTCAGACATCCTGGCCCAGGGCTTTGGCTCCCT
TGGCCTGATGACGTCCGTCCTGGTCTGCCCTGATGGGAAGACGATTGAGGCTGAGGCCGCTCATGGGACC
GTCACCCGCCACTATCGGGAGCACCAGAAGGGCCGGCCCACCAGCACCAACCCCATCGCCAGCATCTTTG
CCTGGACACGTGGCCTGGAGCACCGGGGGAAGCTGGATGGGAACCAAGACCTCATCAGGTTTGCCCAGAT
GCTGGAGAAGGTGTGCGTGGAGACGGTGGAGAGTGGAGCCATGACCAAGGACCTGGCGGGCTGCATTCAC
GGCCTCAGCAATGTGAAGCTGAACGAGCACTTCCTGAACACCACGGACTTCCTCGACACCATCAAGAGCA
ACCTGGACAGAGCCCTGGGCAGGCAGTAG

CDS mutation c.515G>A (Substitution, position 515, G➞A)

**Translation (452 aa):**

>IDH2
MAGYLRVVRSLCRASGSRPAWAPAALTAPTSQEQPRRHYADKRIKVAKPVVEMDGDEMTRIIWQFIKEKL
ILPHVDIQLKYFDLGLPNRDQTDDQVTIDSALATQKYSVAVKCATITPDEARVEEFKLKKMWKSPNGTIR
NILGGTVFREPIICKNIPRLVPGWTKPITIG**K**HAHGDQYKATDFVADRAGTFKMVFTPKDGSGVKEWEVY
NFPAGGVGMGMYNTDESISGFAHSCFQYAIQKKWPLYMSTKNTILKAYDGRFKDIFQEIFDKHYKTDFDK
NKIWYEHRLIDDMVAQVLKSSGGFVWACKNYDGDVQSDILAQGFGSLGLMTSVLVCPDGKTIEAEAAHGT
VTRHYREHQKGRPTSTNPIASIFAWTRGLEHRGKLDGNQDLIRFAQMLEKVCVETVESGAMTKDLAGCIH
GLSNVKLNEHFLNTTDFLDTIKSNLDRALGRQ

AA mutation p.R172K (Substitution - Missense, position 172, R➞K)

GENE *NRAS*

***WILD TYPE DNA NRAS***

***NRAS (ENST00000369535.5)***

**Nucleotide Sequence (570 nt):**

>NRAS
ATGACTGAGTACAAACTGGTGGTGGTTGGAGCAGGTGGTGTTGGGAAAAGCGCACTGACAATCCAGCTAA
TCCAGAACCACTTTGTAGATGAATATGATCCCACCATAGAGGATTCTTACAGAAAACAAGTGGTTATAGA
TGGTGAAACCTGTTTGTTGGACATACTGGATACAGCTGGACAAGAAGAGTACAGTGCCATGAGAGACCAA
TACATGAGGACAGGCGAAGGCTTCCTCTGTGTATTTGCCATCAATAATAGCAAGTCATTTGCGGATATTA
ACCTCTACAGGGAGCAGATTAAGCGAGTAAAAGACTCGGATGATGTACCTATGGTGCTAGTGGGAAACAA
GTGTGATTTGCCAACAAGGACAGTTGATACAAAACAAGCCCACGAACTGGCCAAGAGTTACGGGATTCCA
TTCATTGAAACCTCAGCCAAGACCAGACAGGGTGTTGAAGATGCTTTTTACACACTGGTAAGAGAAATAC
GCCAGTACCGAATGAAAAAACTCAACAGCAGTGATGATGGGACTCAGGGTTGTATGGGATTGCCATGTGT
GGTGATGTAA

**Translation (189 aa):**

>NRAS
MTEYKLVVVGAGGVGKSALTIQLIQNHFVDEYDPTIEDSYRKQVVIDGETCLLDILDTAGQEEYSAMRDQ
YMRTGEGFLCVFAINNSKSFADINLYREQIKRVKDSDDVPMVLVGNKCDLPTRTVDTKQAHELAKSYGIP
FIETSAKTRQGVEDAFYTLVREIRQYRMKKLNSSDDGTQGCMGLPCVVM

***TUMOR***

**Nucleotide Sequence (570 nt):**

>NRAS
ATGACTGAGTACAAACTGGTGGTGGTTGGAGCAGGTGGTGTTGGGAAAAGCGCACTGACAATCCAGCTAA
TCCAGAACCACTTT**A**TAGATGAATATGATCCCACCATAGAGGATTCTTACAGAAAACAAGTGGTTATAGA
TGGTGAAACCTGTTTGTTGGACATACTGGATACAGCTGGACAAGAAGAGTACAGTGCCATGAGAGACCAA
TACATGAGGACAGGCGAAGGCTTCCTCTGTGTATTTGCCATCAATAATAGCAAGTCATTTGCGGATATTA
ACCTCTACAGGGAGCAGATTAAGCGAGTAAAAGACTCGGATGATGTACCTATGGTGCTAGTGGGAAACAA
GTGTGATTTGCCAACAAGGACAGTTGATACAAAACAAGCCCACGAACTGGCCAAGAGTTACGGGATTCCA
TTCATTGAAACCTCAGCCAAGACCAGACAGGGTGTTGAAGATGCTTTTTACACACTGGTAAGAGAAATAC
GCCAGTACCGAATGAAAAAACTCAACAGCAGTGATGATGGGACTCAGGGTTGTATGGGATTGCCATGTGT
GGTGATGTAA

CDS mutation c.85G>A (Substitution, position 85, G➞A)

**Translation (189 aa):**

>NRAS
MTEYKLVVVGAGGVGKSALTIQLIQNHF**I**DEYDPTIEDSYRKQVVIDGETCLLDILDTAGQEEYSAMRDQ
YMRTGEGFLCVFAINNSKSFADINLYREQIKRVKDSDDVPMVLVGNKCDLPTRTVDTKQAHELAKSYGIP
FIETSAKTRQGVEDAFYTLVREIRQYRMKKLNSSDDGTQGCMGLPCVVM

AA mutation p.V29I (Substitution - Missense, position 29, V➞I)

**Polyphen2:**

NRAS:



IDH2:



**cBioPortal:**

IDH2:


Si hacemos click en annotations nos indica que hay un fármaco dirigido contra esta mutación para el tratamiento de pacientes con AML.