*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)

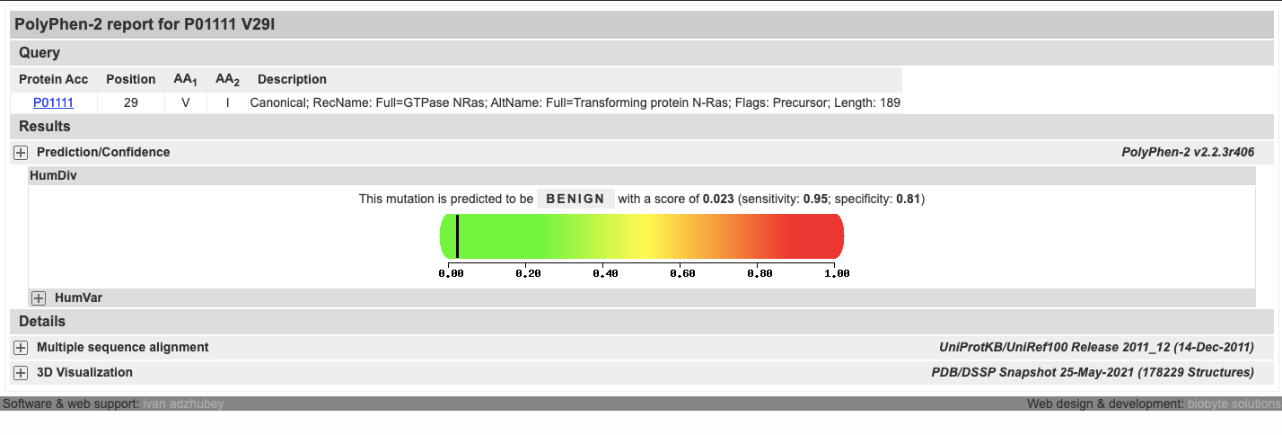
**Translation (189 aa):**

>NRAS  
MTEYKLVVVGAGGVGKSALTIQLIQNHF**I**DEYDPTIEDSYRKQVVIDGETCLLDILDTAGQEEYSAMRDQ  
YMRTGEGFLCVFAINNSKSFADINLYREQIKRVKDSDDVPMVLVGNKCDLPTRTVDTKQAHELAKSYGIP  
FIETSAKTRQGVEDAFYTLVREIRQYRMKKLNSSDDGTQGCMGLPCVVM

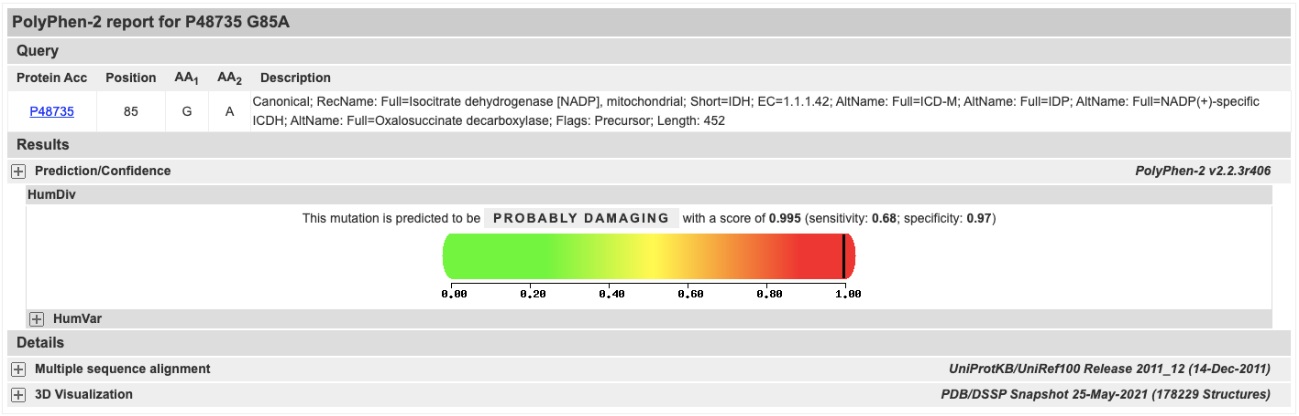
AA mutation p.V29I (Substitution - Missense, position 29, V➞I)

**Polyphen2:**

NRAS:

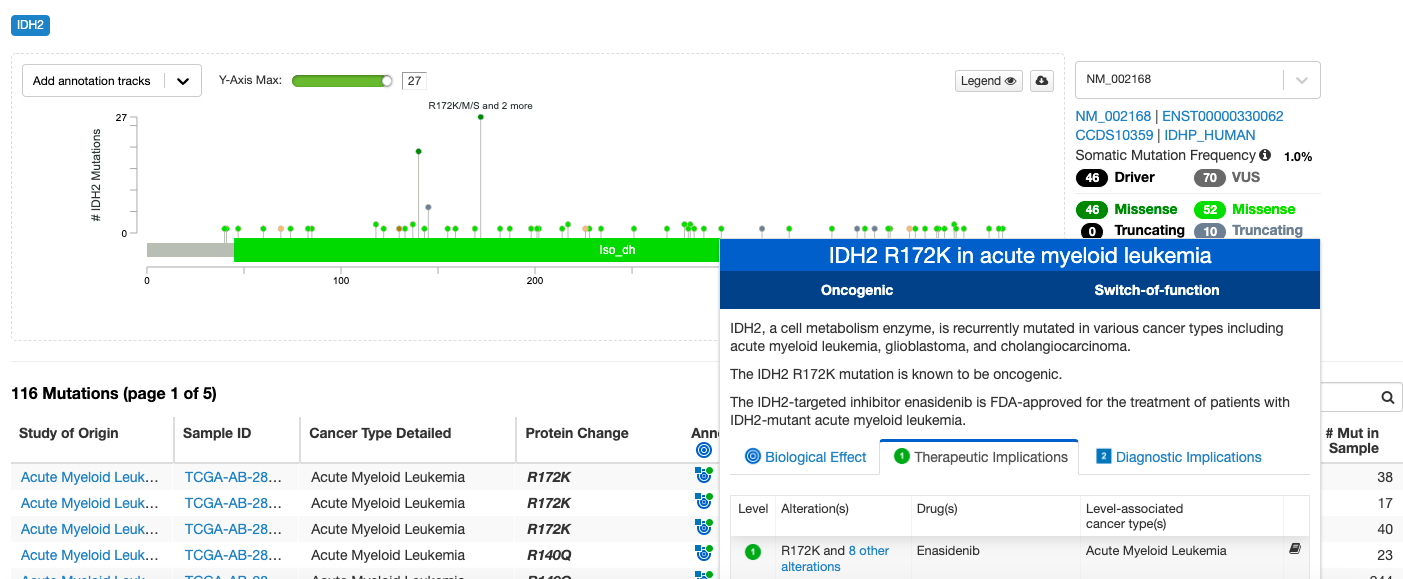


IDH2:



**cBioPortal:**

IDH2:

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