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# Product datasheet for RC403431

## FGFR1 (NM_023110) Human Mutant ORF Clone

### Product data:

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|---|---|
| **Product Type:** | Mutant ORF Clones |
| **Product Name:** | FGFR1 (NM_023110) Human Mutant ORF Clone |
| **Mutation Description:** | Y730X |
| **Affected Codon#:** | 730 |
| **Affected NT#:** | 2190 |
| **Nucleotide Mutation:** | FGFR1 Mutant (Y730X), Myc-DDK-tagged ORF clone of Homo sapiens fibroblast growth factor receptor 1 (FGFR1), transcript variant 1 as transfection-ready DNA |
| **Effect:** | Kallmann syndrome |
| **Symbol:** | FGFR1 |
| **Synonyms:** | bFGF-R-1; BFGFR; CD331; CEK; ECCL; FGFBR; FGFR-1; FLG; FLT-2; FLT2; HBGFR; HH2; HRTFDS; KAL2; N-SAM; OGD |
| **E. coli Selection:** | Kanamycin (25 ug/mL) |
| **Mammalian Cell Selection:** | Neomycin |
| **Vector:** | pCMV6-Entry (PS100001) |
| **Tag:** | Myc-DDK |
| **ACCN:** | NM_023110 |
| **ORF Size:** | 2187 bp |
| **Restriction Sites:** | SgfI-MluI |

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**ORF Nucleotide Sequence:**

>RC403431 representing NM_023110
Red=Cloning site Blue=ORF Green=Tags(s)

TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC
GCCGCGATCGCC

ATGTGGAGCTGGAAGTGCCTCCTCTTCTGGGCTGTGCTGGTCACAGCCACACTCTGCACCGCTAGGCCGT
CCCCGACCTTGCCTGAACAAGCCCAGCCCTGGGGAGCCCCTGTGGAAGTGGAGTCCTTCCTGGTCCACCC
CGGTGACCTGCTGCAGCTTCGCTGTCGGCTGCGGGACGATGTGCAGAGCATCAACTGGCTGCGGGACGGG
GTGCAGCTGGCGGAAAGCAACCGCACCCGCATCACAGGGGAGGAGGTGGAGGTGCAGGACTCCGTGCCCG
CAGACTCCGGCCTCTATGCTTGCGTAACCAGCAGCCCCTCGGGCAGTGACACCACCTACTTCTCCGTCAA
TGTTTCAGATGCTCTCCCCTCCTCGGAGGATGATGATGATGATGATGACTCCTCTTCAGAGGAGAAAGAA
ACAGATAACACCAAACCAAACCGTATGCCCGTAGCTCCATATTGGACATCCCCAGAAAAGATGGAAAAGA
AATTGCATGCAGTGCCGGCTGCCAAGACAGTGAAGTTCAAATGCCCTTCCAGTGGGACCCCAAACCCCAC
ACTGCGCTGGTTGAAAAATGGCAAAGAATTCAAACCTGACCACAGAATTGGAGGCTACAAGGTCCGTTAT
GCCACCTGGAGCATCATAATGGACTCTGTGGTGCCCTCTGACAAGGGCAACTACACCTGCATTGTGGAGA
ATGAGTACGGCAGCATCAACCACACATACCAGCTGGATGTCGTGGAGCGGTCCCCTCACCGGCCCATCCT
GCAAGCAGGGTTGCCCGCCAACAAAACAGTGGCCCTGGGTAGCAACGTGGAGTTCATGTGTAAGGTGTAC
AGTGACCCGCAGCCGCACATCCAGTGGCTAAAGCACATCGAGGTGAATGGGAGCAAGATTGGCCCAGACA
ACCTGCCTTATGTCCAGATCTTGAAGACTGCTGGAGTTAATACCACCGACAAAGAGATGGAGGTGCTTCA
CTTAAGAAATGTCTCCTTTGAGGACGCAGGGGAGTATACGTGCTTGGCGGGTAACTCTATCGGACTCTCC
CATCACTCTGCATGGTTGACCGTTCTGGAAGCCCTGGAAGAGAGGCCGGCAGTGATGACCTCGCCCCTGT
ACCTGGAGATCATCATCTATTGCACAGGGGCCTTCCTCATCTCCTGCATGGTGGGGTCGGTCATCGTCTA
CAAGATGAAGAGTGGTACCAAGAAGAGTGACTTCCACAGCCAGATGGCTGTGCACAAGCTGGCCAAGAGC
ATCCCTCTGCGCAGACAGGTAACAGTGTCTGCTGACTCCAGTGCATCCATGAACTCTGGGGTTCTTCTGG
TTCGGCCATCACGGCTCTCCTCCAGTGGGACTCCCATGCTAGCAGGGGTCTCTGAGTATGAGCTTCCCGA
AGACCCTCGCTGGGAGCTGCCTCGGGACAGACTGGTCTTAGGCAAACCCCTGGGAGAGGGCTGCTTTGGG
CAGGTGGTGTTGGCAGAGGCTATCGGGCTGGACAAGGACAAACCCAACCGTGTGACCAAAGTGGCTGTGA
AGATGTTGAAGTCGGACGCAACAGAGAAAGACTTGTCAGACCTGATCTCAGAAATGGAGATGATGAAGAT
GATCGGGAAGCATAAGAATATCATCAACCTGCTGGGGGCCTGCACGCAGGATGGTCCCTTGTATGTCATC
GTGGAGTATGCCTCCAAGGGCAACCTGCGGGAGTACCTGCAGGCCCGGAGGCCCCCAGGGCTGGAATACT
GCTACAACCCCAGCCACAACCCAGAGGAGCAGCTCTCCTCCAAGGACCTGGTGTCCTGCGCCTACCAGGT
GGCCCGAGGCATGGAGTATCTGGCCTCCAAGAAGTGCATACACCGAGACCTGGCAGCCAGGAATGTCCTG
GTGACAGAGGACAATGTGATGAAGATAGCAGACTTTGGCCTCGCACGGGACATTCACCACATCGACTACT
ATAAAAAGACAACCAACGGCCGACTGCCTGTGAAGTGGATGGCACCCGAGGCATTATTTGACCGGATCTA
CACCCACCAGAGTGATGTGTGGTCTTTCGGGGTGCTCCTGTGGGAGATCTTCACTCTGGGCGGCTCCCCA
TACCCCGGTGTGCCTGTGGAGGAACTTTTCAAGCTGCTGAAGGAGGGTCACCGCATGGACAAGCCCAGTA
ACTGCACCAACGAGCTG

AGCGGACCGACGCGTACGCGGCCGCTCGAGCAGAAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCC
TGGATTACAAGGATGACGACGA TAAGGTTTAA

**Protein Sequence:**

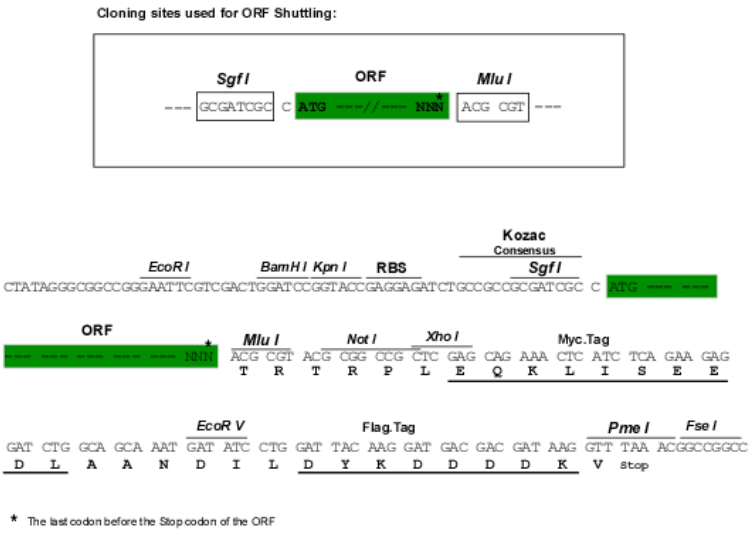
>RC403431 representing NM_023110
Red=Cloning site Green=Tags(s)

MWSWKCLLFWAVLVTATLCTARPSPTLPEQAQPWGAPVEVESFLVHPGDLLQLRCRLRDDVQSINWLRDG
VQLAESNRTRITGEEVEVQDSVPADSGLYACVTSSPSGSDTTYFSVNVSDALPSSEDDDDDDDSSSEEKE
TDNTKPNRMPVAPYWTSPEKMEKKLHAVPAAKTVKFKCPSSGTPNPTLRWLKNGKEFKPDHRIGGYKVRY
ATWSIIMDSVVPSDKGNYTCIVENEYGSINHTYQLDVVERSPHRPILQAGLPANKTVALGSNVEFMCKVY
SDPQPHIQWLKHIEVNGSKIGPDNLPYVQILKTAGVNTTDKEMEVLHLRNVSFEDAGEYTCLAGNSIGLS
HHSAWLTVLEALEERPAVMTSPLYLEIIIYCTGAFLISCMVGSVIVYKMKSGTKKSDFHSQMAVHKLAKS
IPLRRQVTVSADSSASMNSGVLLVRPSRLSSSGTPMLAGVSEYELPEDPRWELPRDRLVLGKPLGEGCFG
QVVLAEAIGLDKDKPNRVTKVAVKMLKSDATEKDLSDLISEMEMMKMIGKHKNIINLLGACTQDGPLYVI
VEYASKGNLREYLQARRPPGLEYCYNPSHNPEEQLSSKDLVSCAYQVARGMEYLASKKCIHRDLAARNVL
VTEDNVMKIADFGLARDIHHIDYYKKTTNGRLPVKWMAPEALFDRIYTHQSDVWSFGVLLWEIFTLGGSP
YPGVPVEELFKLLKEGHRMDKPSNCTNEL

SGPTRTRRLEQKLISEEDLAANDILDYKDDDDKV

**Restriction Sites:** SgfI-MluI

**Cloning Scheme:**



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|---|---|
| **OTI Disclaimer:** | Due to the inherent nature of this plasmid, standard methods to replicate additional amounts of DNA in E. coli are highly likely to result in mutations and/or rearrangements. Therefore, OriGene does not guarantee the capability to replicate this plasmid DNA. Additional amounts of DNA can be purchased from OriGene with batch-specific, full-sequence verification at a reduced cost. Please contact our customer care team at custsupport@origene.com or by calling 301.340.3188 option 3 for pricing and delivery.<br><br>The molecular sequence of this clone aligns with the gene accession number as a point of reference only. However, individual transcript sequences of the same gene can differ through naturally occurring variations (e.g. polymorphisms), each with its own valid existence. This clone is substantially in agreement with the reference, but a complete review of all prevailing variants is recommended prior to use. More info |
| **OTI Annotation:** | This clone was engineered to express the complete ORF with an expression tag. Expression varies depending on the nature of the gene. |
| **Components:** | The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water). |
| **RefSeq:** | NP_075598 |
| **RefSeq Size:** | 2187 bp |
| **RefSeq ORF:** | 2469 bp |
| **Locus ID:** | 2260 |
| **Cytogenetics:** | 8p11.23 |
| **Domains:** | ig, IGc2, IG |
| **Protein Families:** | Druggable Genome, Protein Kinase, Transmembrane |
| **Protein Pathways:** | Adherens junction, MAPK signaling pathway, Melanoma, Pathways in cancer, Prostate cancer, Regulation of actin cytoskeleton |
| **MW:** | 80.2 kDa |

**Gene Summary:** The protein encoded by this gene is a member of the fibroblast growth factor receptor (FGFR) family, where amino acid sequence is highly conserved between members and throughout evolution. FGFR family members differ from one another in their ligand affinities and tissue distribution. A full-length representative protein consists of an extracellular region, composed of three immunoglobulin-like domains, a single hydrophobic membrane-spanning segment and a cytoplasmic tyrosine kinase domain. The extracellular portion of the protein interacts with fibroblast growth factors, setting in motion a cascade of downstream signals, ultimately influencing mitogenesis and differentiation. This particular family member binds both acidic and basic fibroblast growth factors and is involved in limb induction. Mutations in this gene have been associated with Pfeiffer syndrome, Jackson-Weiss syndrome, Antley-Bixler syndrome, osteoglophonic dysplasia, and autosomal dominant Kallmann syndrome 2. Chromosomal aberrations involving this gene are associated with stem cell myeloproliferative disorder and stem cell leukemia lymphoma syndrome. Alternatively spliced variants which encode different protein isoforms have been described; however, not all variants have been fully characterized. [provided by RefSeq, Jul 2008]