abcam

Product datasheet

Human FGFR3 knockout HEK-293 cell line ab259775

3 Images

Overview

Product name	Human FGFR3 knockout HEK-293 cell line
Parental Cell Line	HEK-293
Organism	Human
Mutation description	Knockout achieved by CRISPR/Cas9; X = 1 bp insertion; Frameshift = 99.28%
Passage number	<20
Knockout validation	Next Generation Sequencing (NGS), Western Blot (WB)
Tested applications	**Suitable for:** Next Generation Sequencing, WB
Biosafety level	2
General notes	**Recommended control:** Human wild-type HEK-293 cell line (**ab259776**). Please note a wild-type cell line is not automatically included with a knockout cell line order, if required please add recommended wild-type cell line at no additional cost using the code WILDTYPE-TMTK1. **Cryopreservation cell medium:** Cell Freezing Medium-DMSO Serum free media, contains 8.7% DMSO in MEM supplemented with methyl cellulose. **Culture medium:** DMEM (High Glucose) + 10% FBS **Initial handling guidelines:** Upon arrival, the vial should be stored in liquid nitrogen vapor phase and not at -80°C. Storage at -80°C may result in loss of viability. 1. Thaw the vial in 37°C water bath for approximately 1-2 minutes. 2. Transfer the cell suspension (0.8 mL) to a 15 mL/50 mL conical sterile polypropylene centrifuge tube containing 8.4 mL pre-warmed culture medium, wash vial with an additional 0.8 mL culture medium (total volume 10 mL) to collect remaining cells, and centrifuge at 201 x g (rcf) for 5 minutes at room temperature. 10 mL represents minimum recommended dilution. 20 mL represents maximum recommended dilution. 3. Resuspend the cell pellet in 5 mL pre-warmed culture medium and count using a haemocytometer or alternative cell counting method. Based on cell count, seed cells in an appropriate cell culture flask at a density of $2x10^4$ cells/cm^2. Seeding density is given as a guide only and should be scaled to align with individual lab schedules. 4. Incubate the culture at 37°C incubator with 5% CO_2. Cultures should be monitored daily. **Subculture guidelines:** All seeding densities should be based on cell counts gained by established methods. A guide seeding density of $2x10^4$ cells/cm^2 is recommended. A partial media change 24 hours prior to subculture may be helpful to encourage growth, if required.

Cells should be passaged when they have achieved 80-90% confluence.
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We will provide viable cells that proliferate on revival.

Properties

Number of cells	1 x 10^6 cells/vial, 1 mL
Adherent /Suspension	Adherent
Tissue	Kidney
Cell type	epithelial
Gender	Female
Mycoplasma free	Yes
Storage instructions	Shipped on Dry Ice. Store in liquid nitrogen.
Storage buffer	Constituents: 8.7% Dimethylsulfoxide, 2% Cellulose, methyl ether

Target

Function	Receptor for acidic and basic fibroblast growth factors. Preferentially binds FGF1.
Tissue specificity	Expressed in brain, kidney and testis. Very low or no expression in spleen, heart, and muscle. In 20- to 22-week old fetuses it is expressed at high level in kidney, lung, small intestine and brain, and to a lower degree in spleen, liver, and muscle. Isoform 2 is detected in epithelial cells. Isoform 1 is not detected in epithelial cells. Isoform 1 and isoform 2 are detected in fibroblastic cells.
Involvement in disease	Defects in FGFR3 are the cause of achondroplasia (ACH) [MIM:100800]. ACH is an autosomal dominant disease and is the most frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. Defects in FGFR3 are the cause of Crouzon syndrome with acanthosis nigricans (CAN) [MIM:612247]. Classic Crouzon disease which is caused by mutations in the FGFR2 gene is characterized by craniosynostosis (premature fusion of the skull sutures), and facial hypoplasia. Crouzon syndrome with acanthosis nigricans (a skin disorder characterized by pigmentation anomalies), CAN, is considered to be an independent disorder from classic Crouzon syndrome. CAN is characterized by additional more severe physical manifestation, such as Chiari malformation, hydrocephalus, and atresia or stenosis of the choanas, and is caused by a specific mutation (Ala-391 to Glu) in the transmembrane domain of FGFR3. It is proposed to have an autosomal dominant mode of inheritance. Defects in FGFR3 are a cause of thanatophoric dysplasia type (TD) [MIM:187600, 187601]; also known as thanatophoric dwarfism or platyspondylic lethal skeletal dysplasia Sand Diego type (PLSD-SD). TD is the most common neonatal lethal skeletal dysplasia. Affected individuals display features similar to those seen in homozygous achondroplasia. It causes severe shortening of the limbs with macrocephaly, narrow thorax and short ribs. In the most common subtype, TD1, femur are curved, while in TD2, straight femurs are associated with cloverleaf skull. Mutations affecting different functional domains of FGFR3 cause different forms of this lethal disorder. Defects in FGFR3 are a cause of hypochondroplasia (HCH) [MIM:146000]. HCH is an autosomal dominant disease and is characterized by disproportionate short stature. It resembles

achondroplasia, but with a less severe phenotype.
Defects in FGFR3 are a cause of susceptibility to bladder cancer (BLC) [MIM:109800]. A malignancy originating in tissues of the urinary bladder. It often presents with multiple tumors appearing at different times and at different sites in the bladder. Most bladder cancers are transitional cell carcinomas. They begin in cells that normally make up the inner lining of the bladder. Other types of bladder cancer include squamous cell carcinoma (cancer that begins in thin, flat cells) and adenocarcinoma (cancer that begins in cells that make and release mucus and other fluids). Bladder cancer is a complex disorder with both genetic and environmental influences. Note=Somatic mutations can constitutively activate FGFR3.
Defects in FGFR3 are a cause of cervical cancer (CERCA) [MIM:603956]. A malignant neoplasm of the cervix, typically originating from a dysplastic or premalignant lesion previously present at the active squamocolumnar junction. The transformation from mild dysplastic to invasive carcinoma generally occurs slowly within several years, although the rate of this process varies widely. Carcinoma in situ is particularly known to precede invasive cervical cancer in most cases. Cervical cancer is strongly associated with infection by oncogenic types of human papillomavirus.
Defects in FGFR3 are the cause of camptodactyly tall stature and hearing loss syndrome (CATSHL syndrome) [MIM:610474]. CATSHL syndrome is an autosomal dominant syndrome characterized by permanent and irreducible flexion of one or more fingers of the hand and/or feet, tall stature, scoliosis and/or a pectus excavatum, and hearing loss. Affected individuals have developmental delay and/or mental retardation, and several of these have microcephaly. Radiographic findings included tall vertebral bodies with irregular borders and broad femoral metaphyses with long tubular shafts. On audiological exam, each tested member have bilateral sensorineural hearing loss and absent otoacoustic emissions. The hearing loss was congenital or developed in early infancy, progressed variably in early childhood, and range from mild to severe. Computed tomography and magnetic resonance imaging reveal that the brain, middle ear, and inner ear are structurally normal.
Defects in FGFR3 are a cause of multiple myeloma (MM) [MIM:254500]. MM is a malignant tumor of plasma cells usually arising in the bone marrow and characterized by diffuse involvement of the skeletal system, hyperglobulinemia, Bence-Jones proteinuria and anemia. Complications of multiple myeloma are bone pain, hypercalcemia, renal failure and spinal cord compression. The aberrant antibodies that are produced lead to impaired humoral immunity and patients have a high prevalence of infection. Amyloidosis may develop in some patients. Multiple myeloma is part of a spectrum of diseases ranging from monoclonal gammopathy of unknown significance (MGUS) to plasma cell leukemia. Note=A chromosomal aberration involving FGFR3 is found in multiple myeloma. Translocation t(4;14)(p16.3;q32.3) with the IgH locus.
Defects in FGFR3 are a cause of lacrimo-auriculo-dento-digital syndrome (LADDS) [MIM:149730]; also known as Levy-Hollister syndrome. LADDS is a form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. LADDS is an autosomal dominant syndrome characterized by aplastic/hypoplastic lacrimal and salivary glands and ducts, cup-shaped ears, hearing loss, hypodontia and enamel hypoplasia, and distal limb segments anomalies. In addition to these cardinal features, facial dysmorphism, malformations of the kidney and respiratory system and abnormal genitalia have been reported. Craniosynostosis and severe syndactyly are not observed.
Defects in FGFR3 are a cause of keratinocytic non-epidermolytic nevus (KNEN) [MIM:162900]; also known as pigmented moles. Epidermal nevi of the common, non-organoid and non-epidermolytic type are benign skin lesions and may vary in their extent from a single (usually linear) lesion to widespread and systematized involvement. They may be present at birth or develop early during childhood.
Defects in FGFR3 are a cause of Muenke syndrome (MNKS) [MIM:602849]; also known as Muenke non-syndromic coronal craniosynostosis. MNKS is a condition characterized by premature closure of coronal suture of skull during development (coronal craniosynostosis), which affects the shape of the head and face. It may be uni- or bilateral. When bilateral, it is

characterized by a skull with a small antero-posterior diameter (brachycephaly), often with a decrease in the depth of the orbits and hypoplasia of the maxillae. Unilateral closure of the coronal sutures leads to flattening of the orbit on the involved side (plagiocephaly). The intellect is normal. In addition to coronal craniosynostosis some affected individuals show skeletal abnormalities of hands and feet, sensorineural hearing loss, mental retardation and respiratory insufficiency. Defects in FGFR3 are a cause of keratosis seborrheic (KERSEB) [MIM:182000]. A common benign skin tumor. Seborrheic keratoses usually begin with the appearance of one or more sharply defined, light brown, flat macules. The lesions may be sparse or numerous. As they initially grow, they develop a velvety to finely verrucous surface, followed by an uneven warty surface with multiple plugged follicles and a dull or lackluster appearance.

Sequence similarities Belongs to the protein kinase superfamily. Tyr protein kinase family. Fibroblast growth factor receptor subfamily.
Contains 3 Ig-like C2-type (immunoglobulin-like) domains.
Contains 1 protein kinase domain.

Cellular localization Membrane.

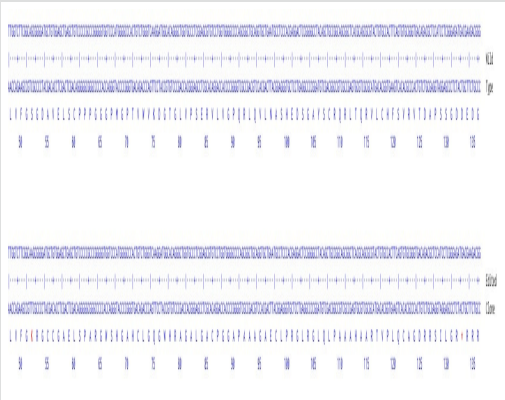
Applications

The Abpromise guarantee Our **Abpromise guarantee** covers the use of ab259775 in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

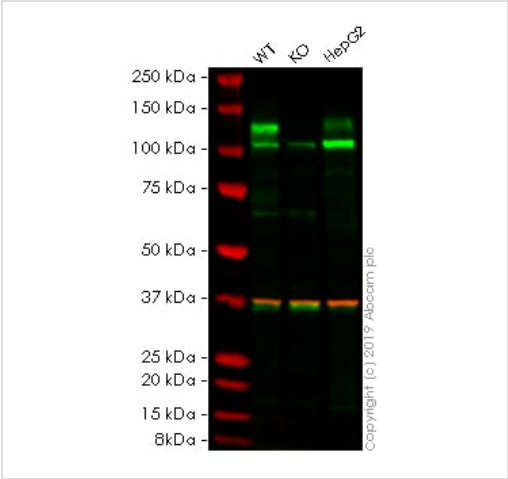
Application	Abreviews	Notes
Next Generation Sequencing		Use at an assay dependent concentration.
WB		Use at an assay dependent concentration.

Images



Next Generation Sequencing - Human FGFR3 knockout HEK-293 cell line (ab259775)

1 bp deletion after Gly51 of the WT protein



Western blot - Human FGFR3 knockout HEK-293 cell line (ab259775)

All lanes : Anti-FGFR3 antibody [EPR2304(3)] (**ab133644**) at 1/1000 dilution

Lane 1 : Wild-type HEK-293 (Human epithelial cell line from embryonic kidney) whole cell lysate
Lane 2 : FGFR3 knockout HEK-293 (Human epithelial cell line from embryonic kidney) whole cell lysate
Lane 3 : Hep G2 (Human liver hepatocellular carcinoma cell line) whole cell lysate

Lysates/proteins at 20 µg per lane.

Performed under reducing conditions.

Lanes 1 - 3: Merged signal (red and green). Green - **ab133644** observed at 125kDa. Red - loading control, **ab9484**, observed at 37 kDa.

ab133644 was shown to recognize in wild-type HEK-293 cells as signal was lost at the expected MW in FGFR3 knockout cell line ab259775 (knockout cell lysate **ab259781**). Additional cross-reactive bands were observed in the wild-type and knockout samples. Wild-type and FGFR3 knockout samples were subjected to SDS-PAGE. Ab133644 and **ab9484** (Mouse anti-GAPDH loading control) were incubated overnight at 4°C at 1/1000 dilution and 1/20000 dilution respectively. Blots were developed with Goat anti-Rabbit IgG H&L (IRDye® 800CW) preabsorbed **ab216773** and Goat anti-Mouse IgG H&L (IRDye® 680RD) preabsorbed **ab216776** secondary antibodies at 1/20000 dilution for 1 hour at room temperature before imaging.

Next Generation Sequencing - Human FGFR3 knockout HEK-293 cell line (ab259775)

Knockout achieved by CRISPR/Cas9; X = 1 bp insertion; Frameshift = 99.28%

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