



## Properties

**Storage instructions** Store at -80°C. Please refer to protocols.

Components	1 kit
ab260139 - Human ASS1 knockout HeLa cell lysate (Lyophilized)	1 x 100µg
ab255552 - Human wild-type HeLa cell lysate (Lyophilized)	1 x 100µg

<b>Cell type</b>	epithelial
<b>Disease</b>	Adenocarcinoma
<b>Gender</b>	Female
<b>STR Analysis</b>	Amelogenin X D5S818: 11, 12 D13S317: 12, 13.3 D7S820: 8, 12 D16S539: 9, 10 vWA: 16, 18 TH01: 7 TPOX: 8, 12 CSF1PO: 9, 10

## Target

**Pathway** Amino-acid biosynthesis; L-arginine biosynthesis; L-arginine from L-ornithine and carbamoyl phosphate: step 2/3.  
Nitrogen metabolism; urea cycle; (N(omega)-L-arginino)succinate from L-aspartate and L-citrulline: step 1/1.

**Involvement in disease** Defects in ASS1 are the cause of citrullinemia type 1 (CTLN1) [MIM:215700]. Citrullinemia belongs to the urea cycle disorders. It is an autosomal recessive disease characterized primarily by elevated serum and urine citrulline levels. Ammonia intoxication is another manifestation. CTLN1 usually manifests in the first few days of life. Affected infants appear normal at birth, but as ammonia builds up in the body they present symptoms such as lethargy, poor feeding, vomiting, seizures and loss of consciousness. Less commonly, a milder CTLN1 form can develop later in childhood or adulthood.

**Sequence similarities** Belongs to the argininosuccinate synthase family. Type 1 subfamily.

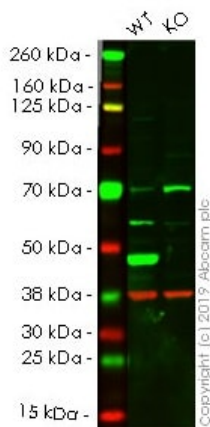
## Applications

Our [Abpromise guarantee](#) covers the use of **ab257143** 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
WB		Use at an assay dependent concentration.

## Images



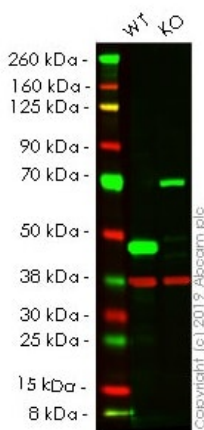
Western blot - Human ASS1 knockout HeLa cell lysate (ab257143)

**Lane 1:** Wild-type HeLa cell lysate (20µg)

**Lane 2:** ASS1 knockout HeLa cell lysate (20µg)

**Lanes 1- 2:** Merged signal (red and green). Green - [ab170952](#) observed at 47 kDa. Red - loading control [ab8245](#) observed at 37 kDa.

[ab170952](#) Anti-ASS1 antibody [EPR12398] was shown to specifically react with ASS1 in wild-type HeLa cells in western blot. Loss of signal was observed when knockout cell line [ab264989](#) (knockout cell lysate ab257143) was used. Wild-type and ASS1 knockout samples were subjected to SDS-PAGE. Membrane was blocked for 1 hour at room temperature in 0.1% TBST with 3% non-fat dried milk. [ab170952](#) and Anti-GAPDH antibody [6C5] - Loading Control ([ab8245](#)) were incubated overnight at 4 °C at 1 in 1000 dilution and 1 in 20000 dilution respectively. Blots were developed with Goat anti-Rabbit IgG H&L (IRDye® 800CW) preadsorbed ([ab216773](#)) and Goat anti-Mouse IgG H&L (IRDye® 680RD) preadsorbed ([ab216776](#)) secondary antibodies at 1 in 20000 dilution for 1 hour at room temperature before imaging.



Western blot - Human ASS1 knockout HeLa cell lysate (ab257143)

**Lane 1:** Wild-type HeLa cell lysate (20µg)

**Lane 2:** ASS1 knockout HeLa cell lysate (20µg)

**Lanes 1- 2:** Merged signal (red and green). Green - [ab170900](#) observed at 47 kDa. Red - loading control [ab8245](#) observed at 37 kDa.

[ab170900](#) Anti-ASS1 antibody [EPR12399(B)] - C-terminal was shown to specifically react with ASS1 in wild-type HeLa cells in western blot. Loss of signal was observed when knockout cell line [ab264989](#) (knockout cell lysate ab257143) was used. Wild-type and ASS1 knockout samples were subjected to SDS-PAGE. Membrane was blocked for 1 hour at room temperature in 0.1% TBST with 3% non-fat dried milk. [ab170900](#) and Anti-GAPDH antibody [6C5] - Loading Control ([ab8245](#)) were incubated overnight at 4 °C at 1 in 1000 dilution and 1 in 20000 dilution respectively. Blots were developed with Goat anti-Rabbit IgG H&L (IRDye® 800CW) preadsorbed ([ab216773](#)) and Goat anti-Mouse IgG H&L (IRDye® 680RD) preadsorbed ([ab216776](#)) secondary

antibodies at 1 in 20000 dilution for 1 hour at room temperature before imaging.

```
Mut  TCCC GCCAGAGCCTATGTCCAGCAAAGGCTCCGTGGTCTGGCCTACAGTGGCGGCCTG
WT   TCCC GCCAGAGCCTATGTCCAGCAAAGGCTCCGTGGTCTGGCCTACAGTGGCGGCCTG
```

Allele-1: 1 bp insertion in exon 3

Sanger Sequencing - Human ASS1 knockout HeLa  
cell lysate (ab257143)

```
Mut  CGCTATGTCCAGCAAAGGCT*****|n s e r t i o n*****CCGTGGTCTGGCCTACAGT
WT   CGCTATGTCCAGCAAAGGCT                CCGTGGTCTGGCCTACAGT
```

Allele-2: Insertion of the selection cassette in exon 3

Sanger Sequencing - Human ASS1 knockout HeLa  
cell lysate (ab257143)

**Please note:** All products are "FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC PROCEDURES"

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