

CLOVES Syndrome and the PI3K/AKT Pathway

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Boston
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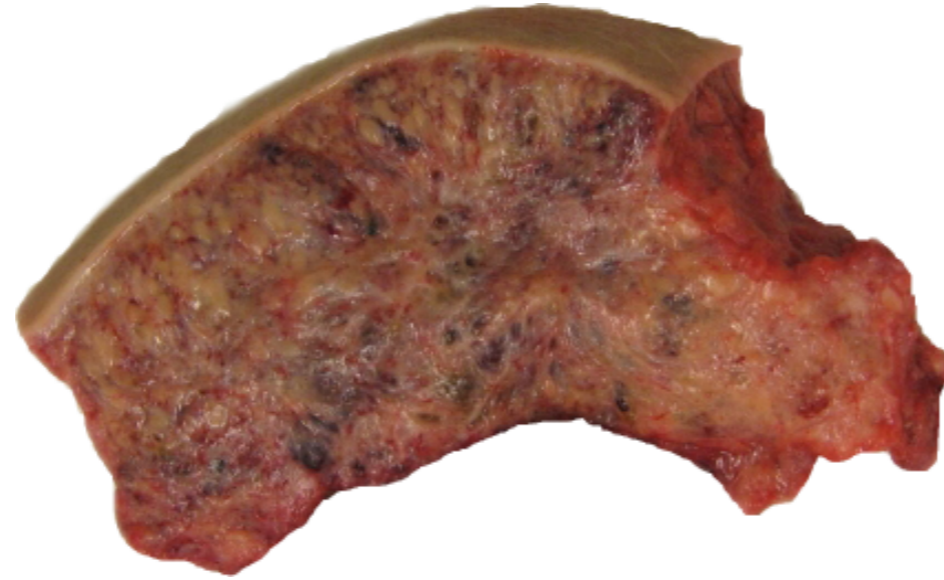
Method to Identify the Genetic Cause of CLOVES Syndrome



Archival Lesional Tissue

Bar-coded Genomic Libraries

Targeted/Exome Sequencing



Fresh or Frozen Lesional Tissue

Bar-coded RNA Libraries

RNA Sequencing

Bar-coded Genomic Libraries

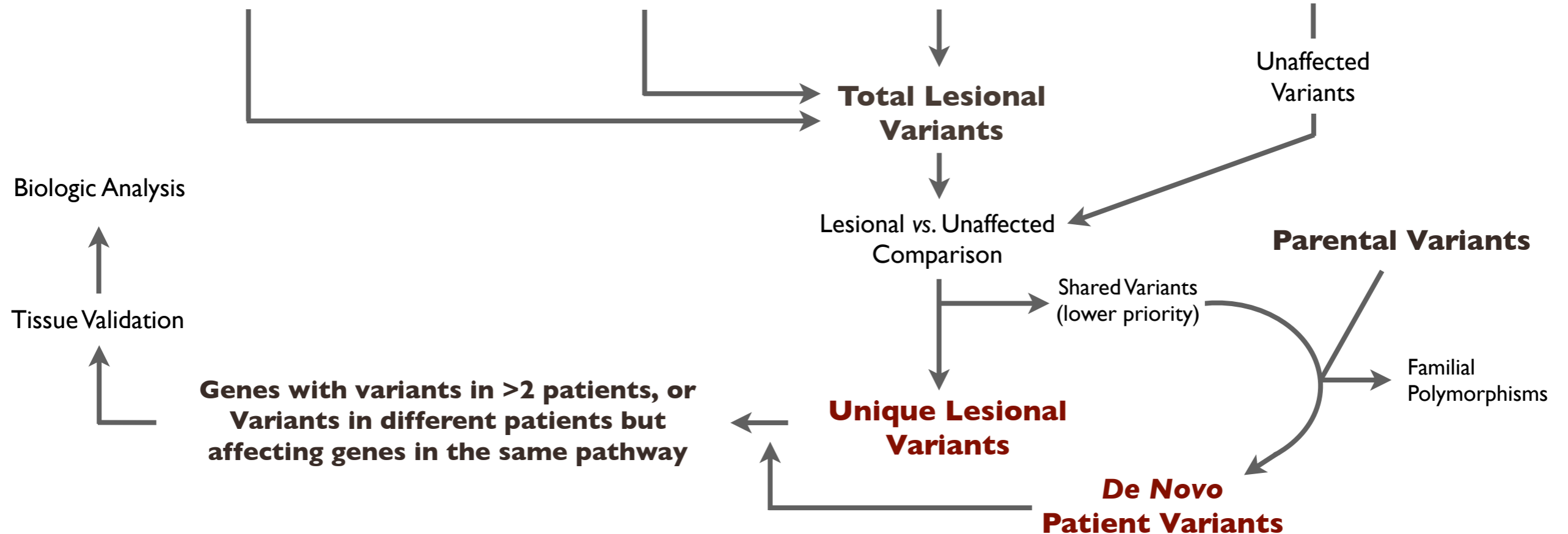
Exome Sequencing



Unaffected Blood

Bar-coded Genomic Libraries

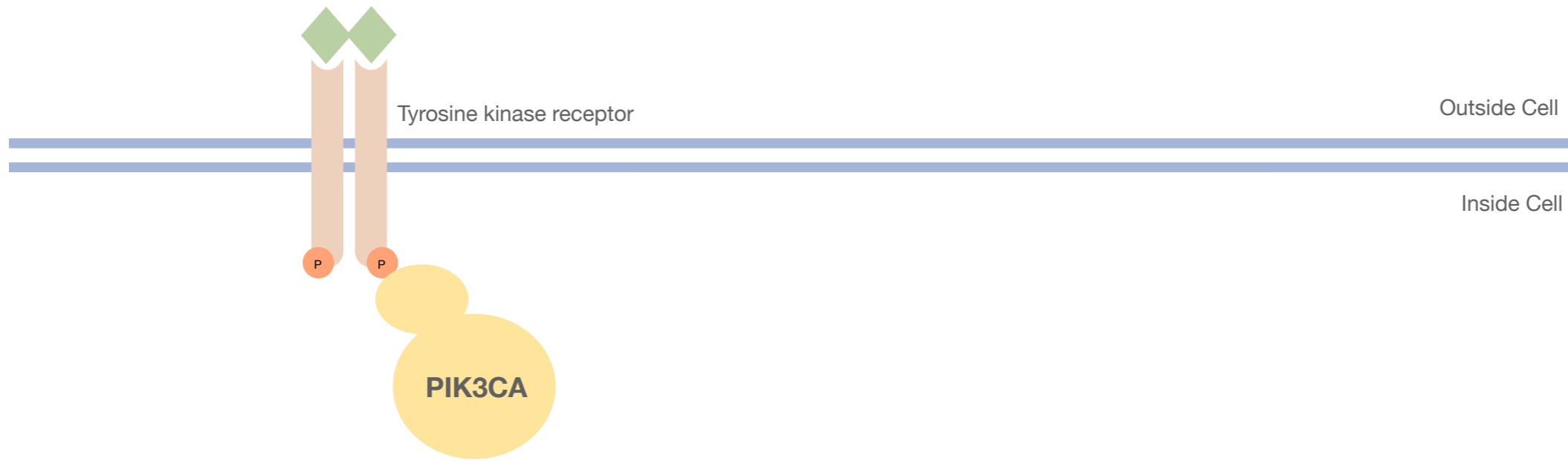
Exome Sequencing



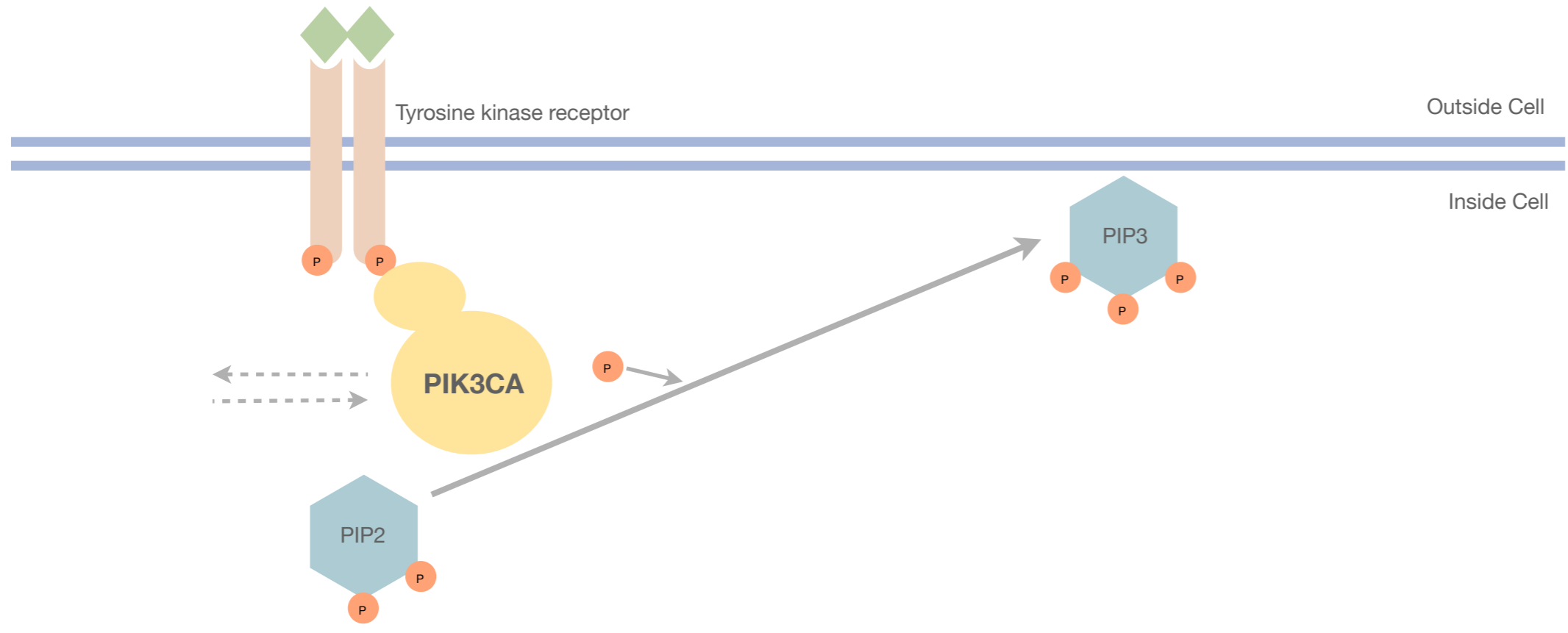
The PI3K/ AKT/ mTOR Pathway



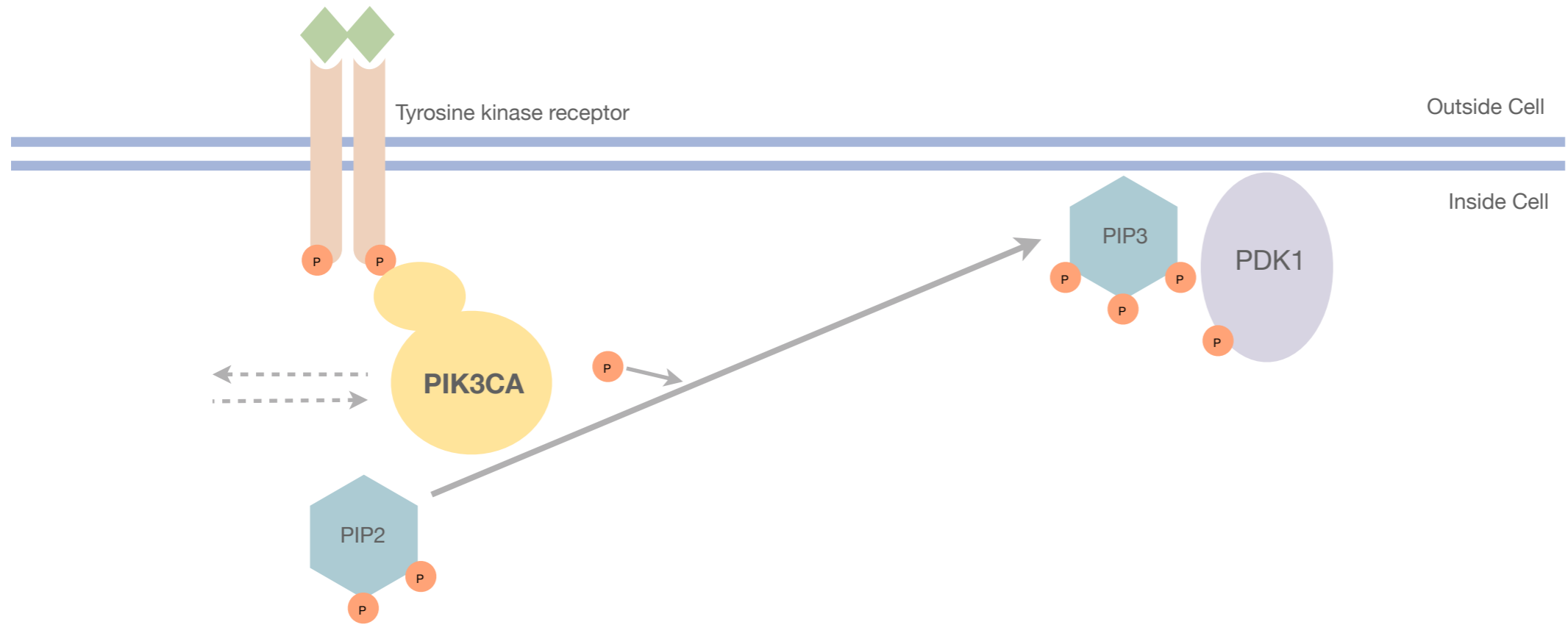
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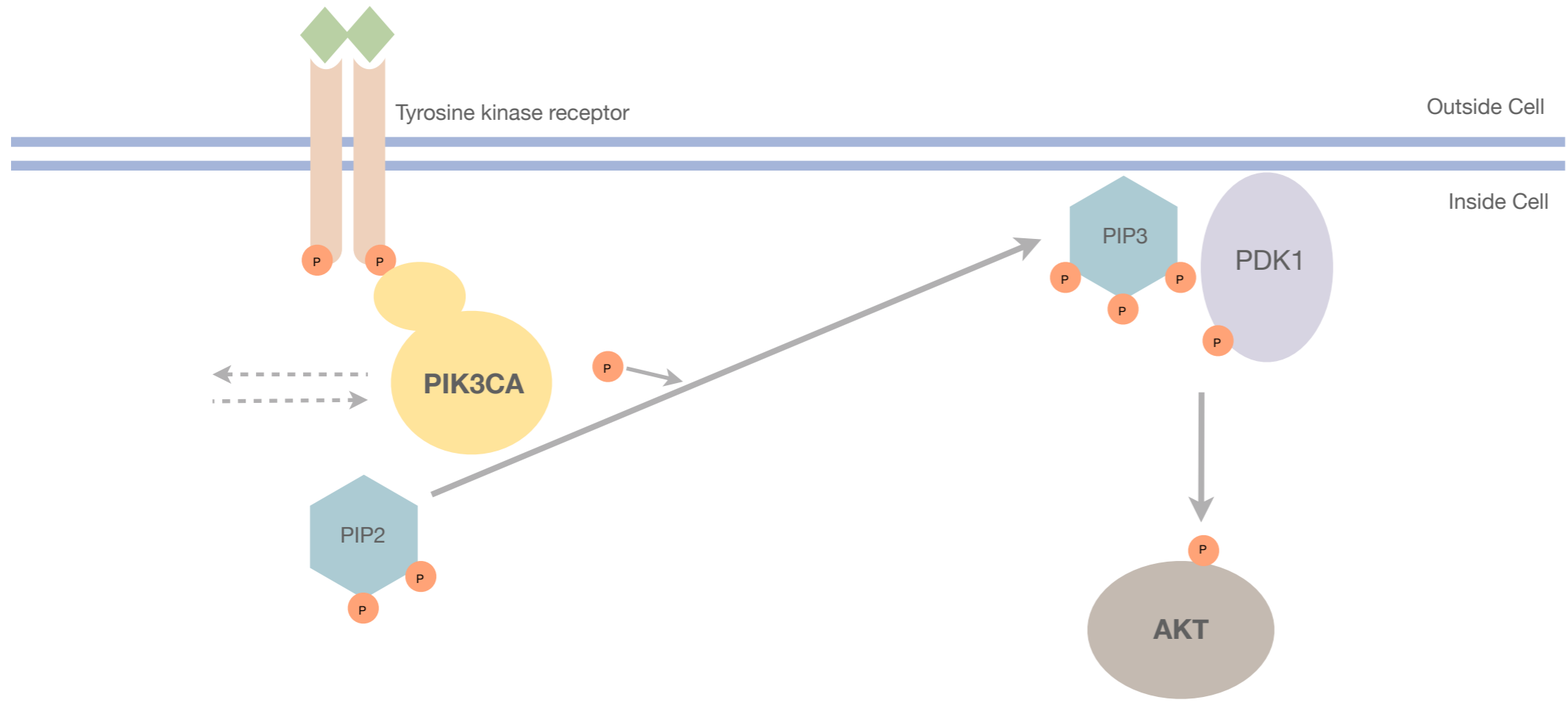
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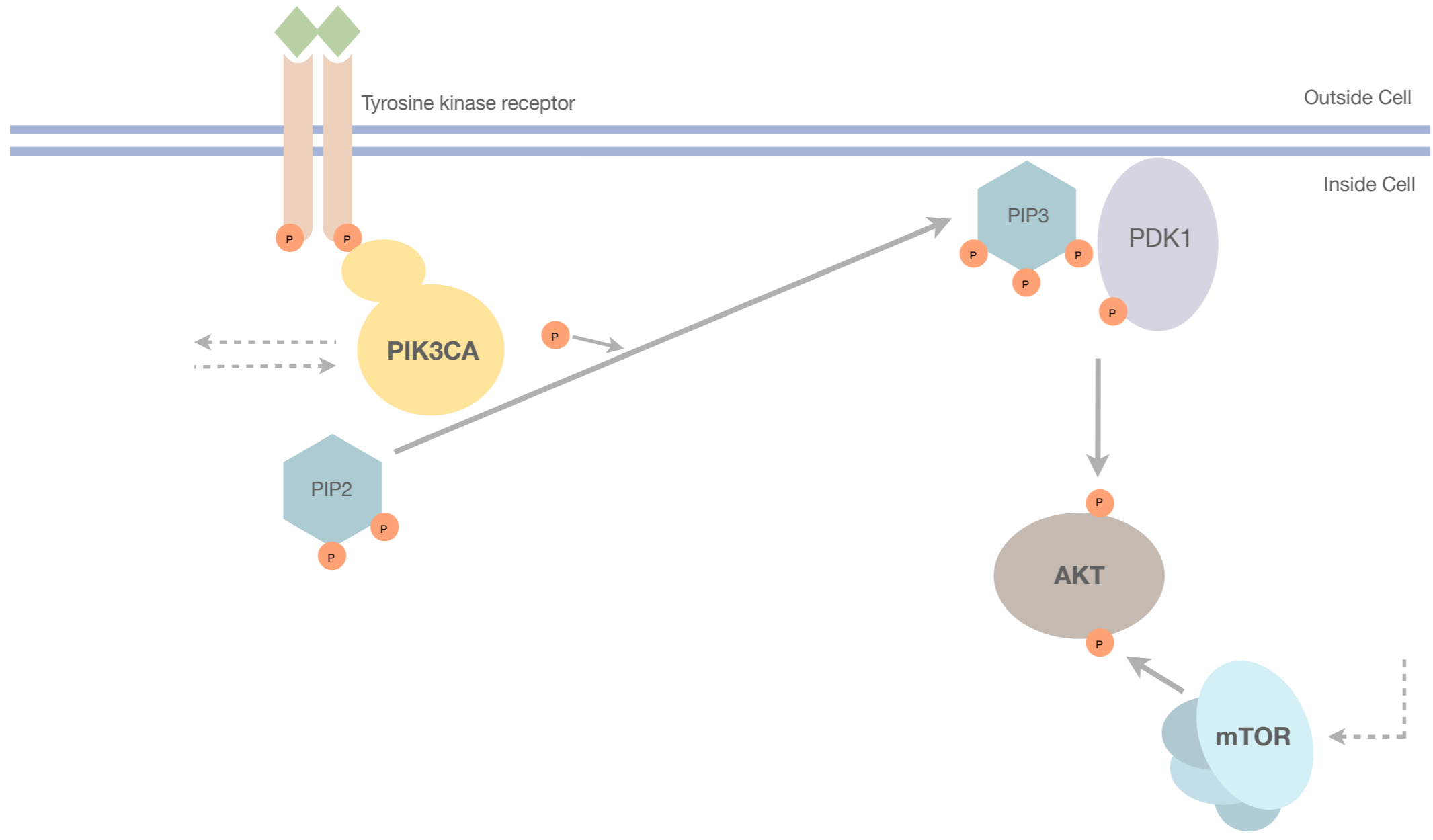
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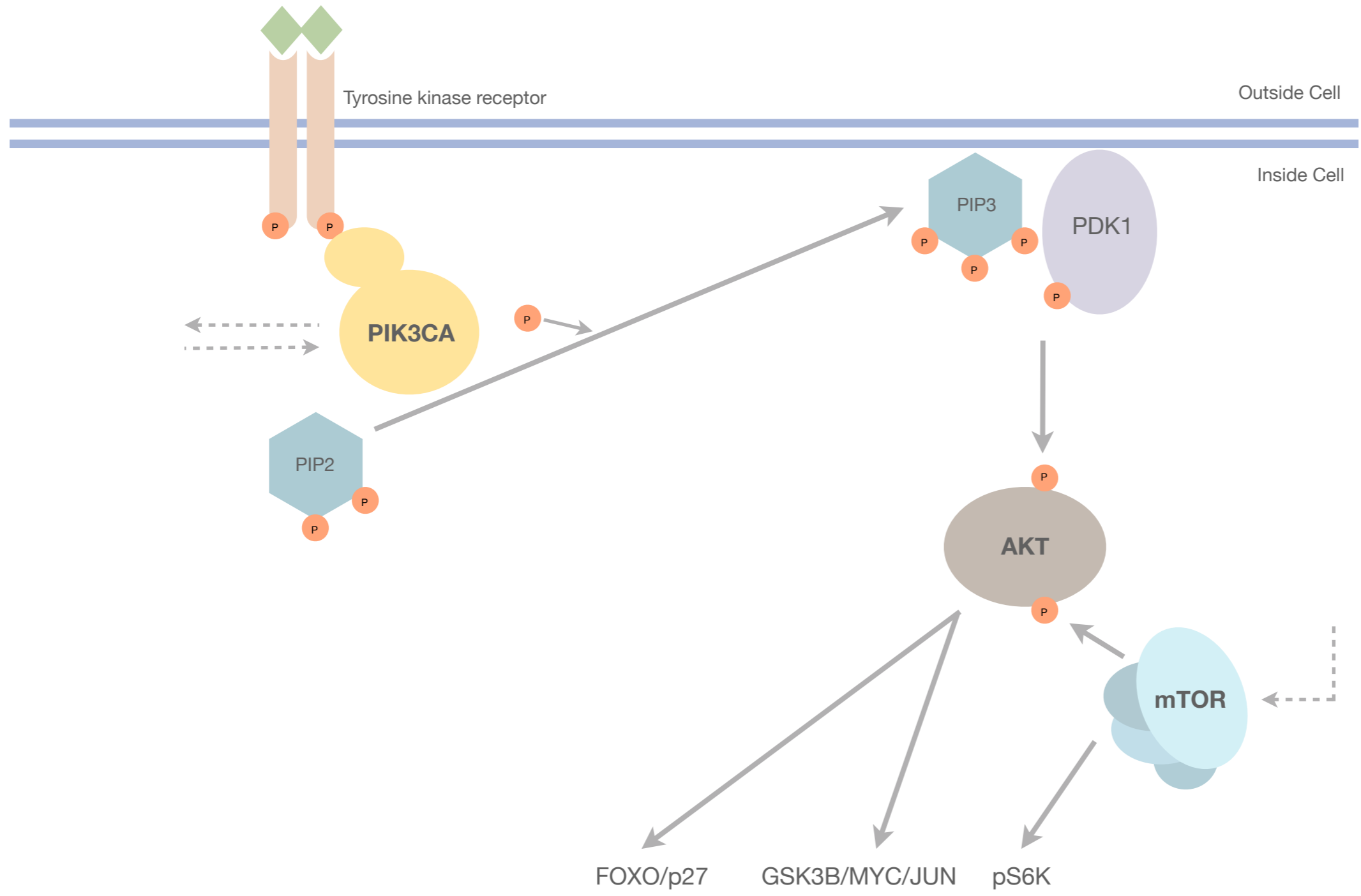
The PI3K/ AKT/ mTOR Pathway



The PI3K/ AKT/ mTOR Pathway

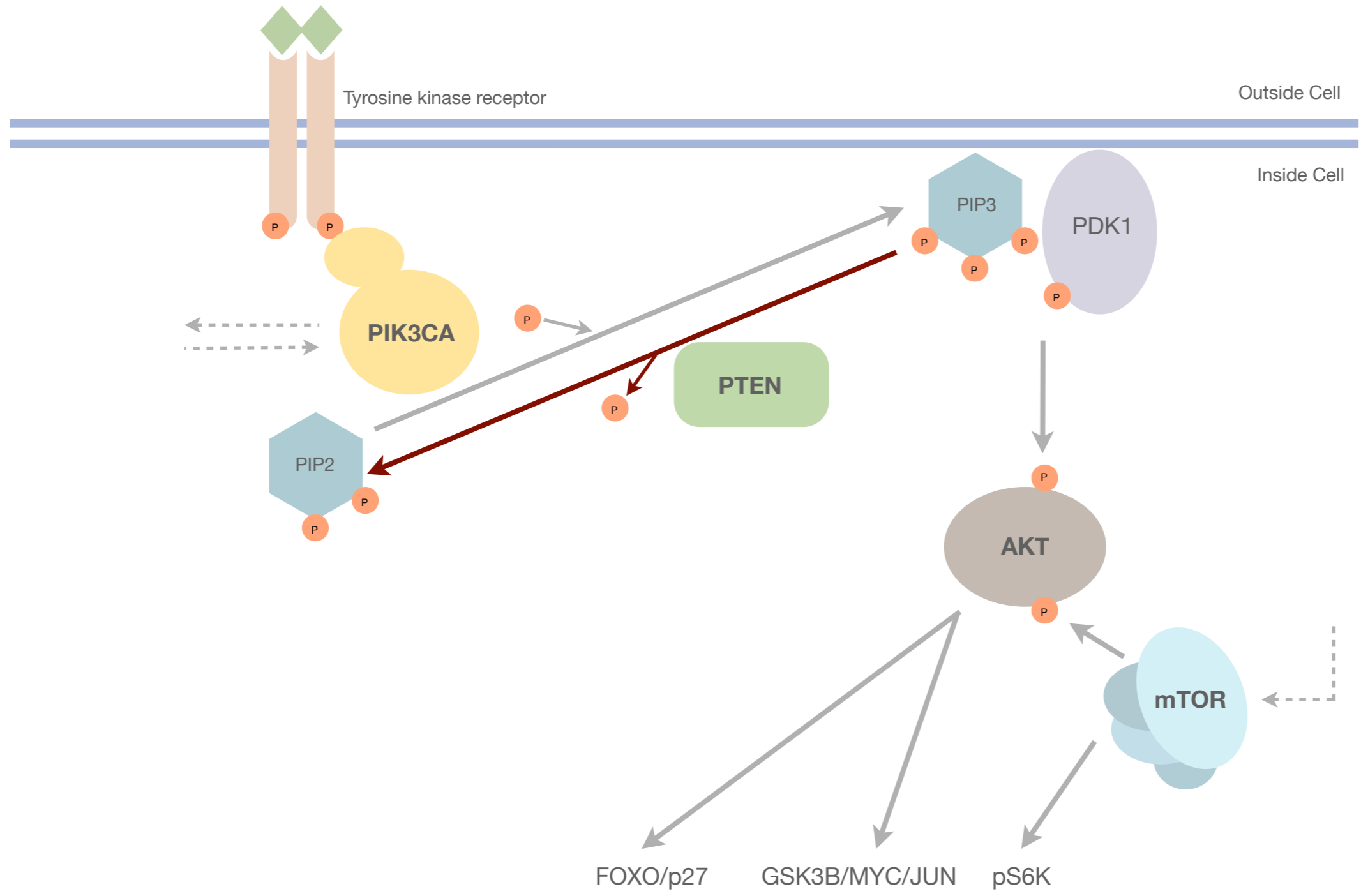


The PI3K/ AKT/ mTOR Pathway



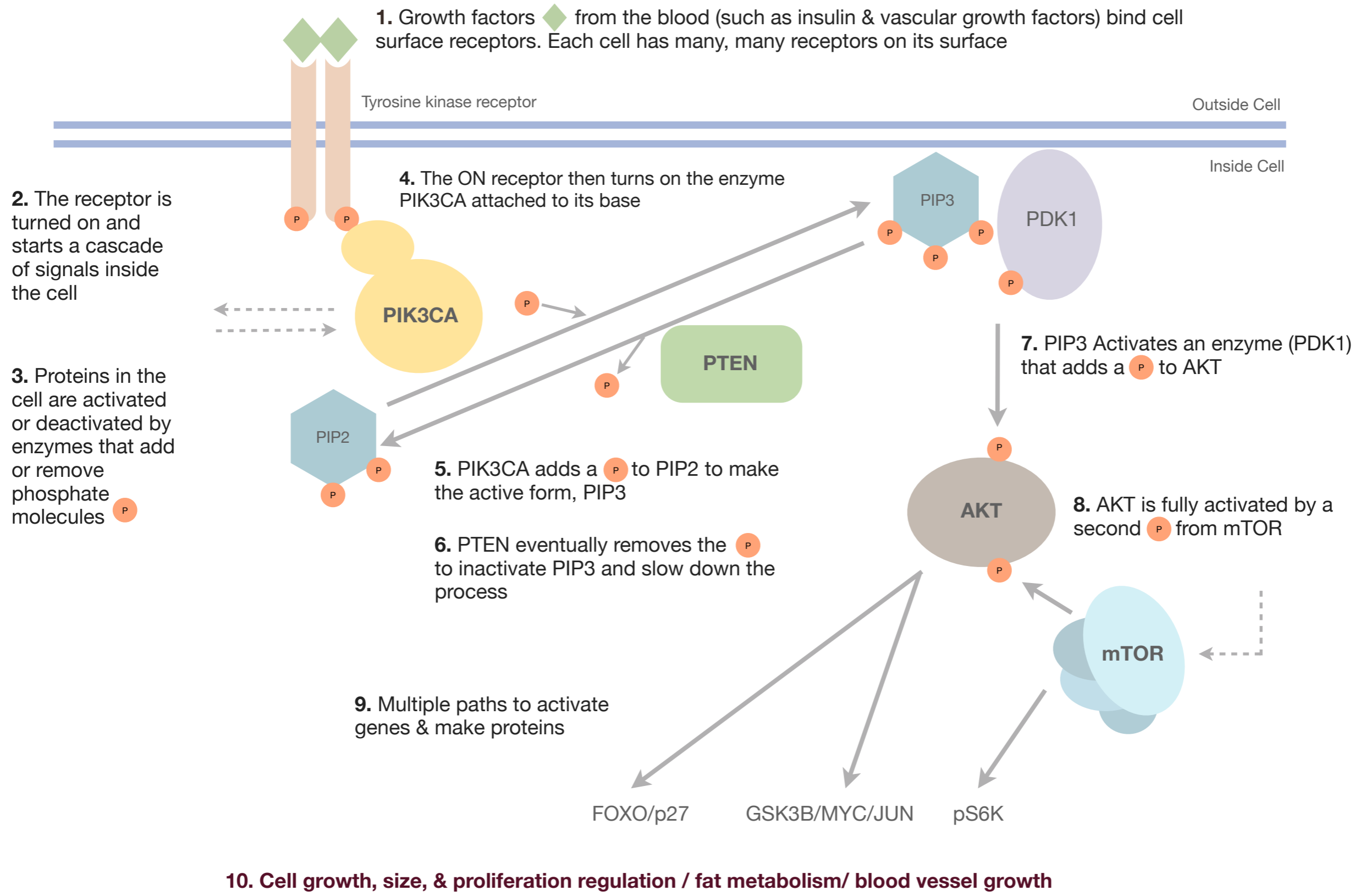
Cell growth, size, & proliferation regulation / fat metabolism/ blood vessel growth

The PI3K/ AKT/ mTOR Pathway

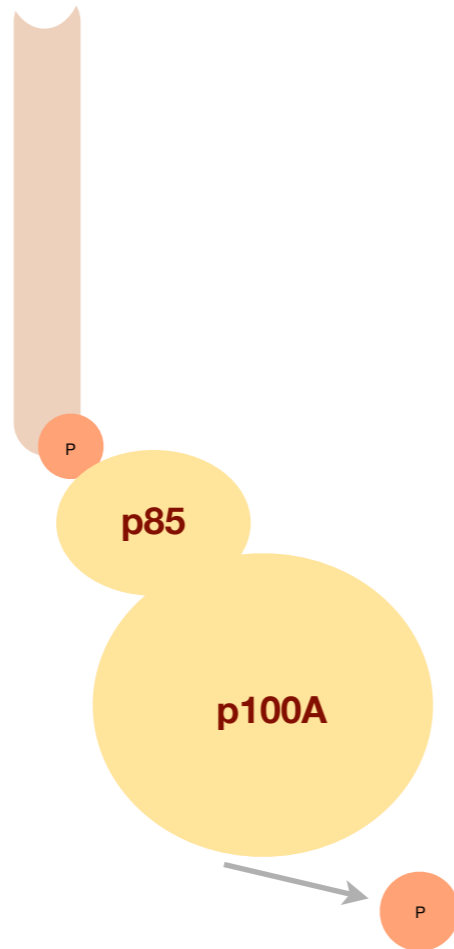


Cell growth, size, & proliferation regulation / fat metabolism/ blood vessel growth

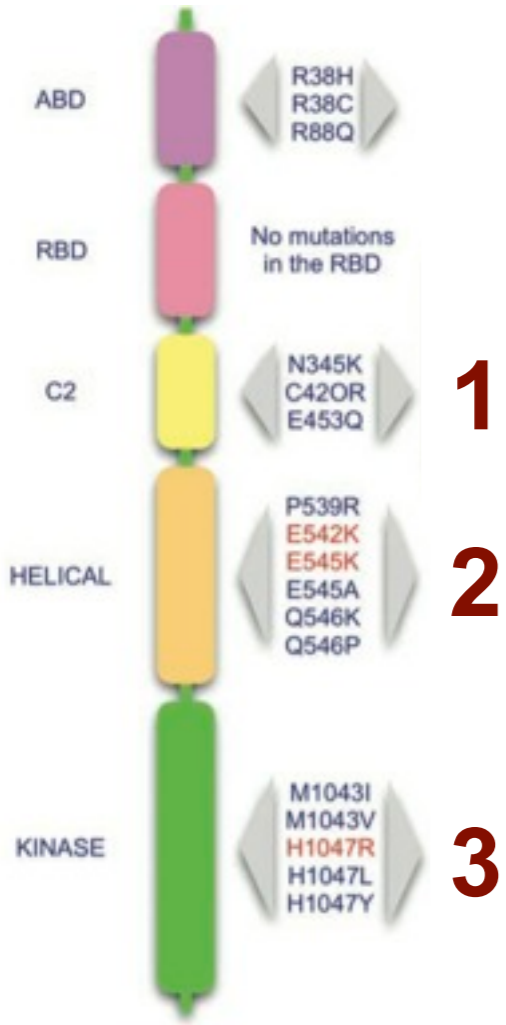
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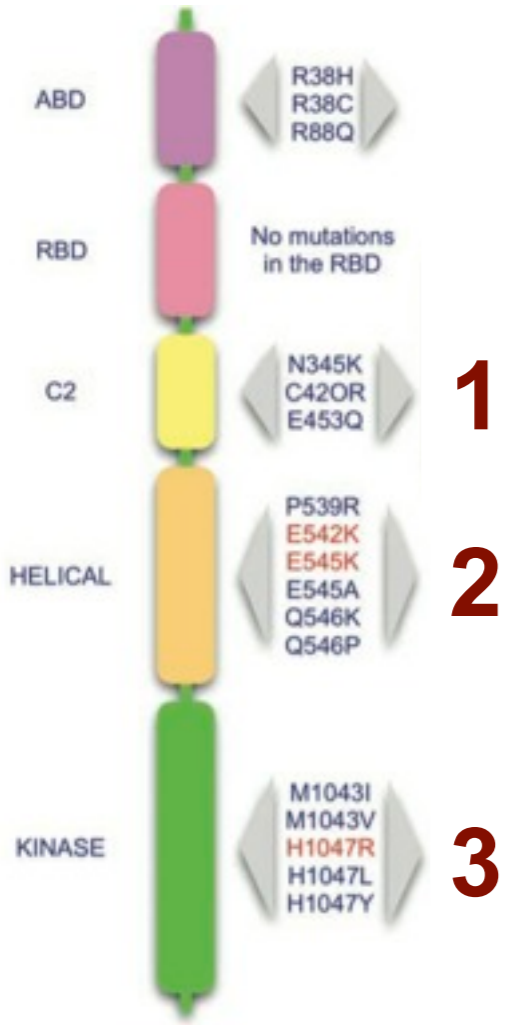
PIK3CA Mutations & CLOVES Syndrome



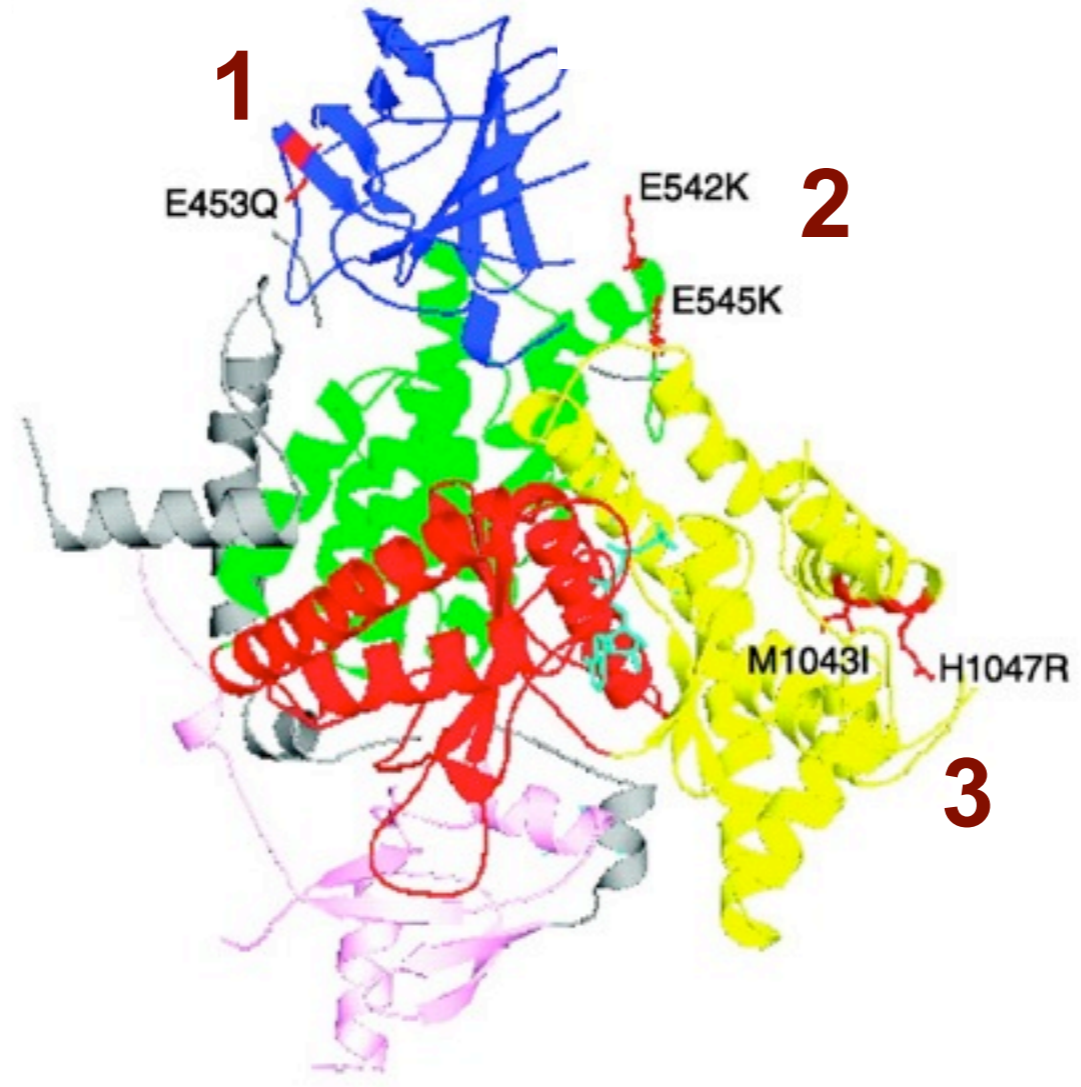
PIK3CA Mutations & CLOVES Syndrome



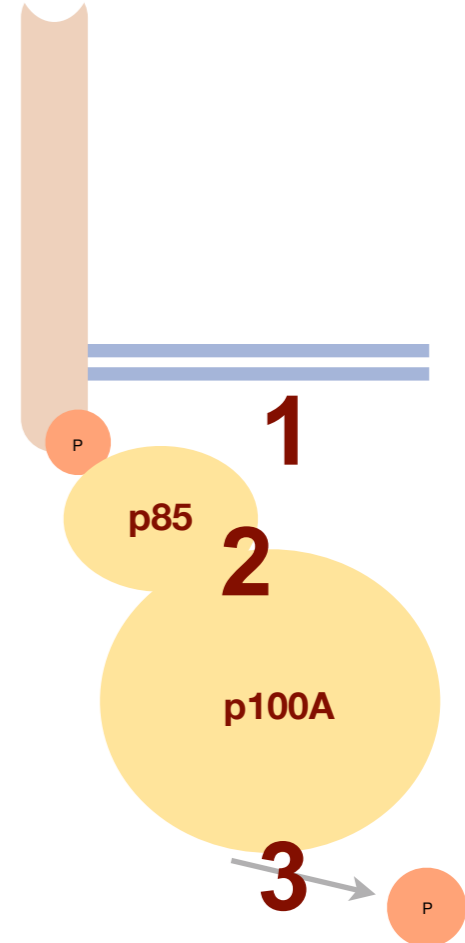
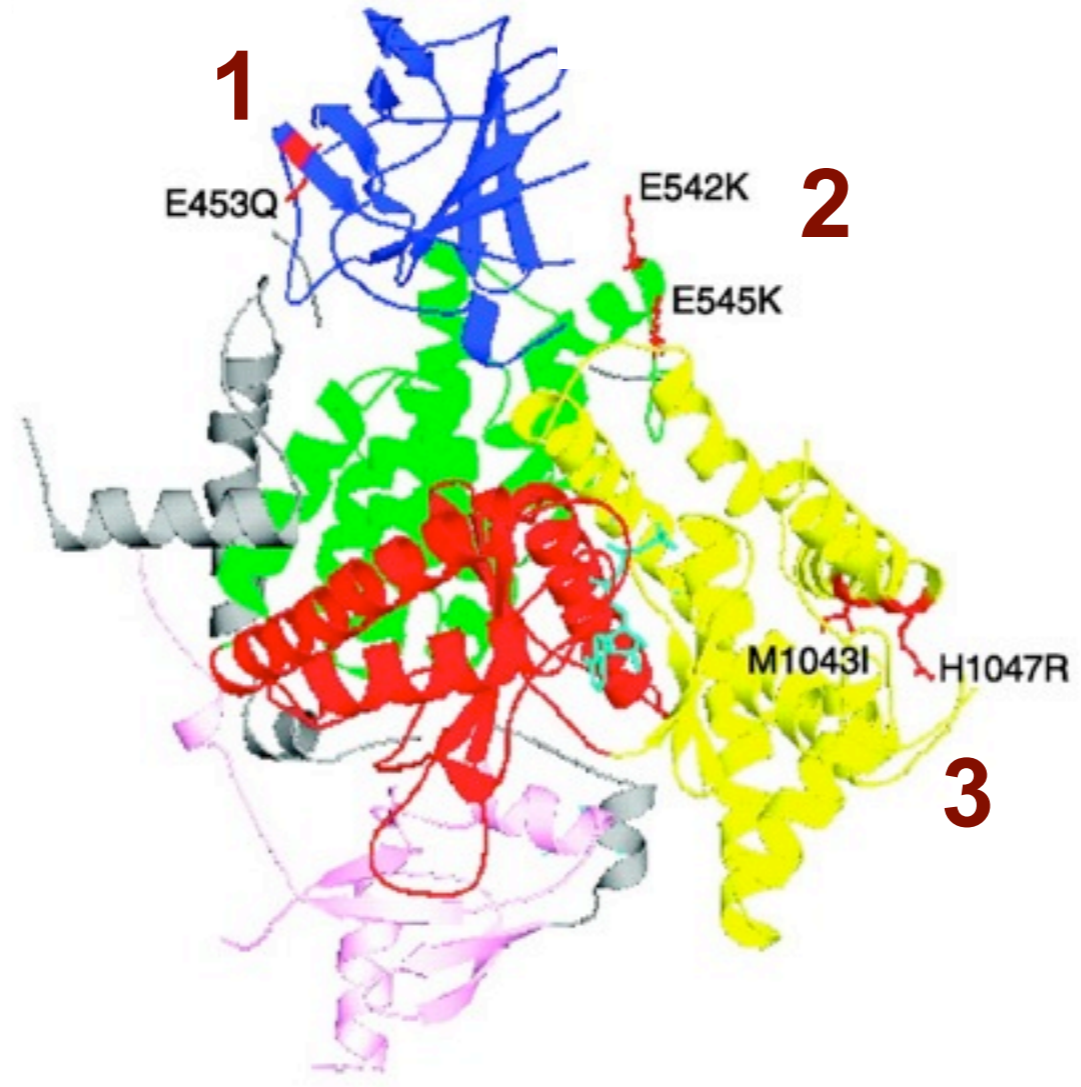
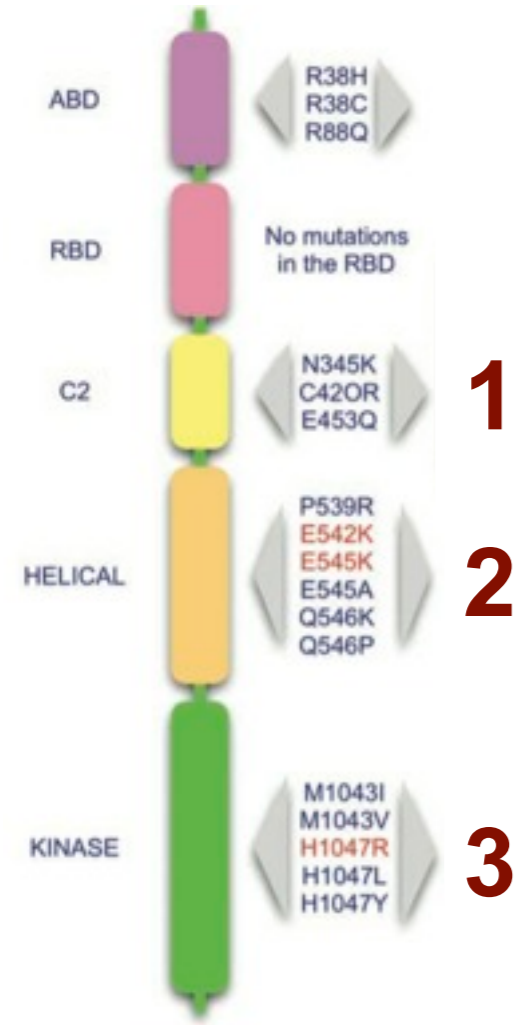
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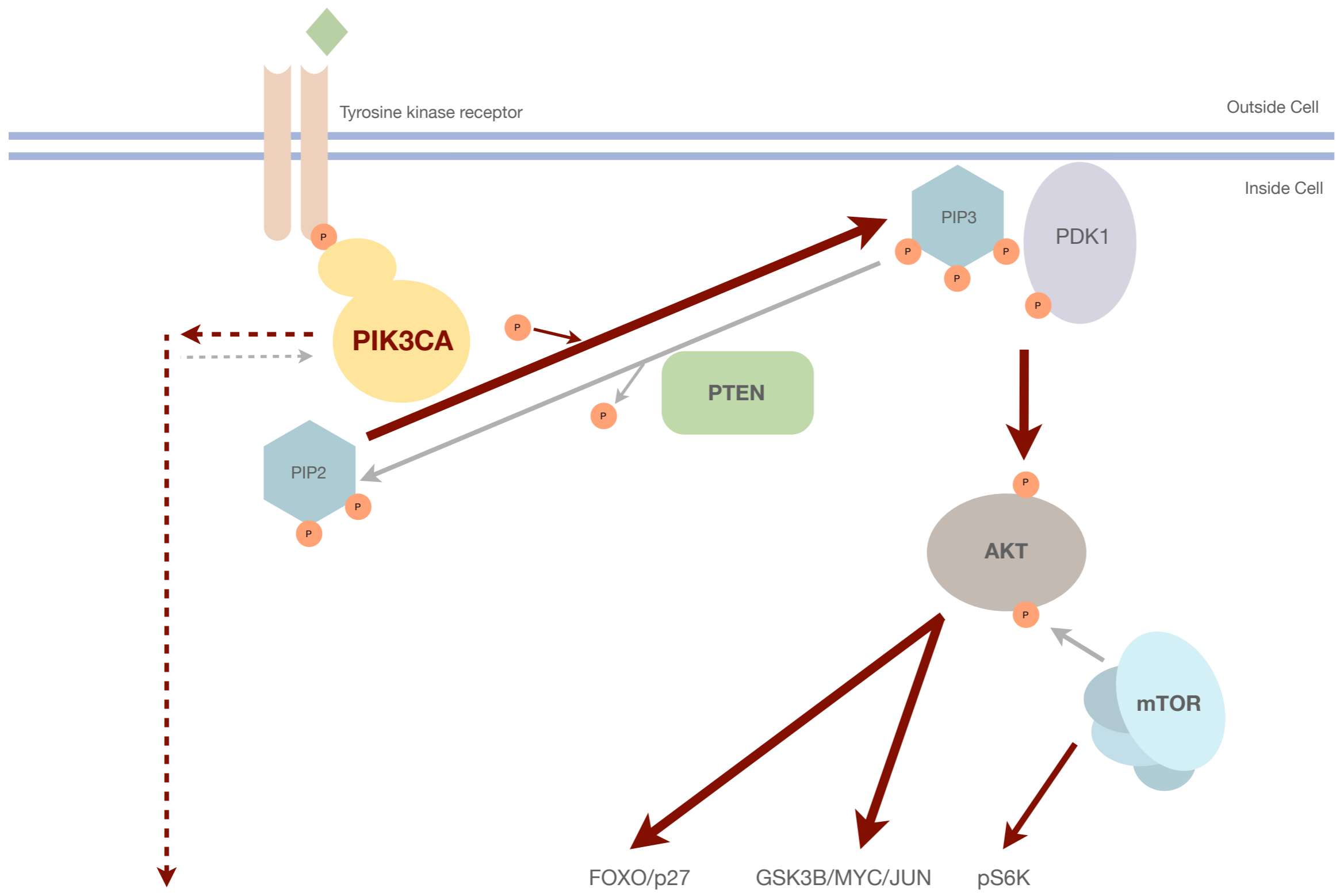
1
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3



PIK3CA Mutations & CLOVES Syndrome

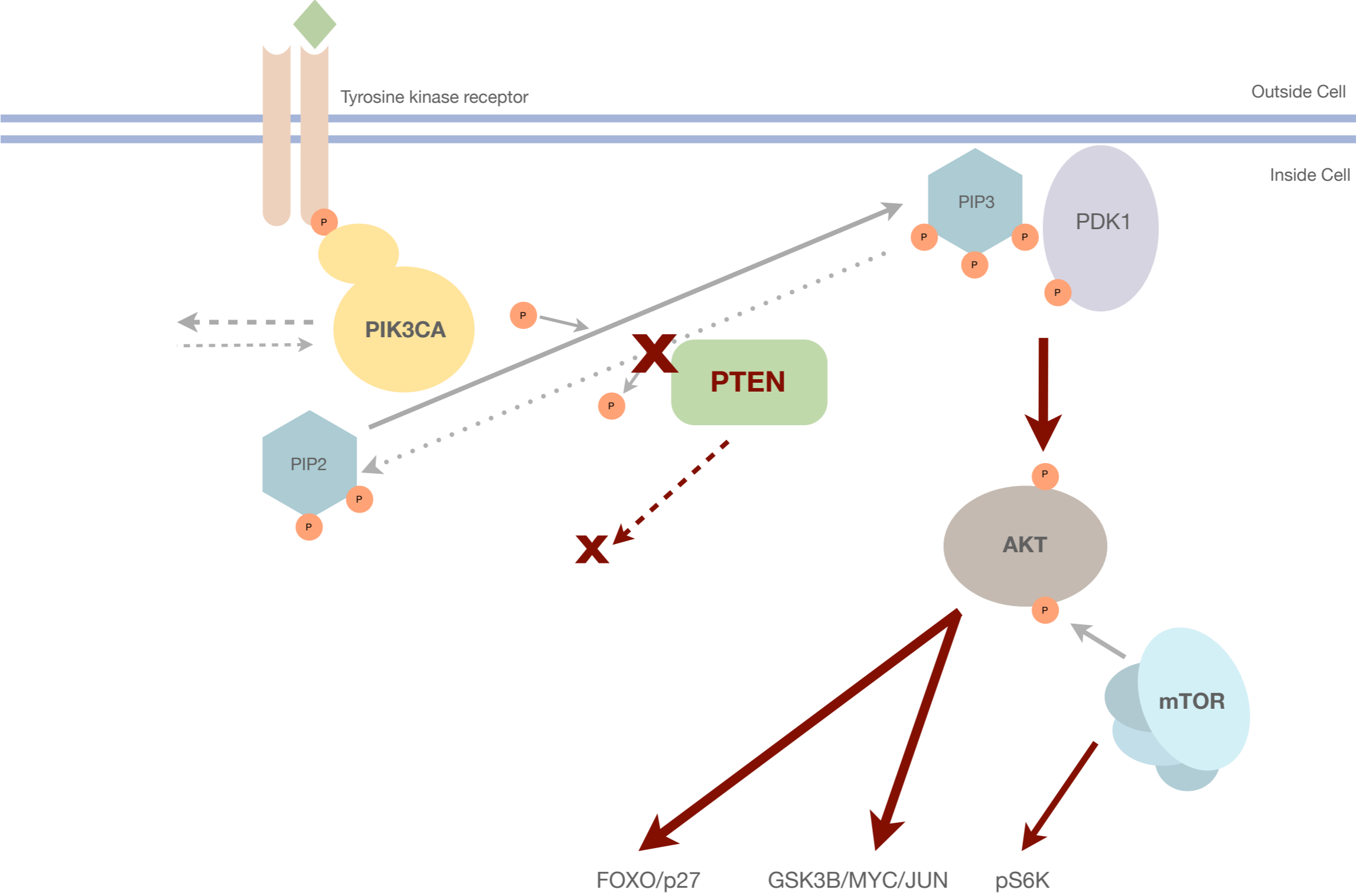


CLOVES Syndrome/ Klippel-Trenaunay Syndrome (PIK3CA) Megalencephaly ± Capillary Malformation/ MPPH (PIK3CA, PIK3R2)



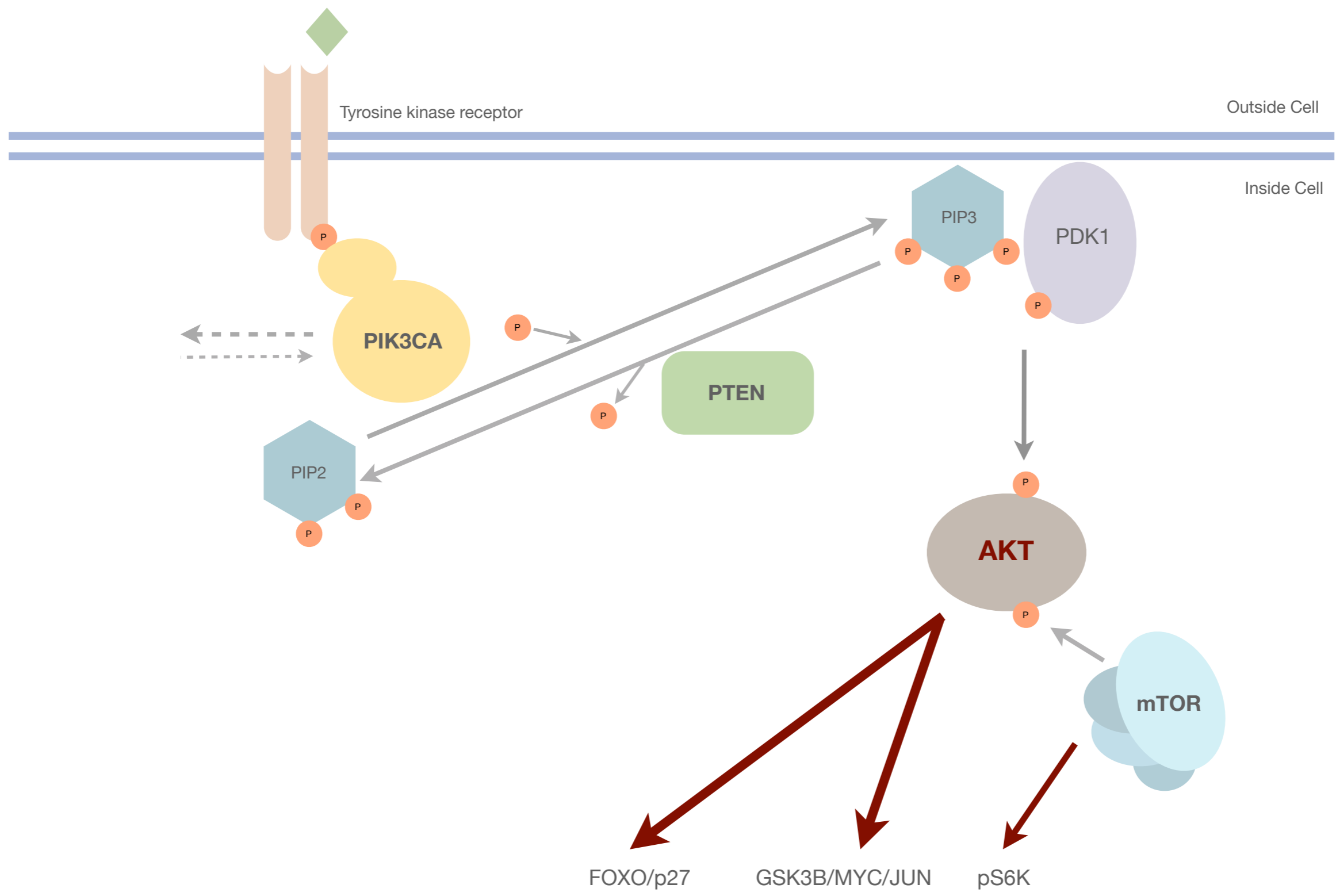
Cell growth, size, & proliferation regulation / fat metabolism/ blood vessel growth

PTEN Hamartoma Tumor Syndrome



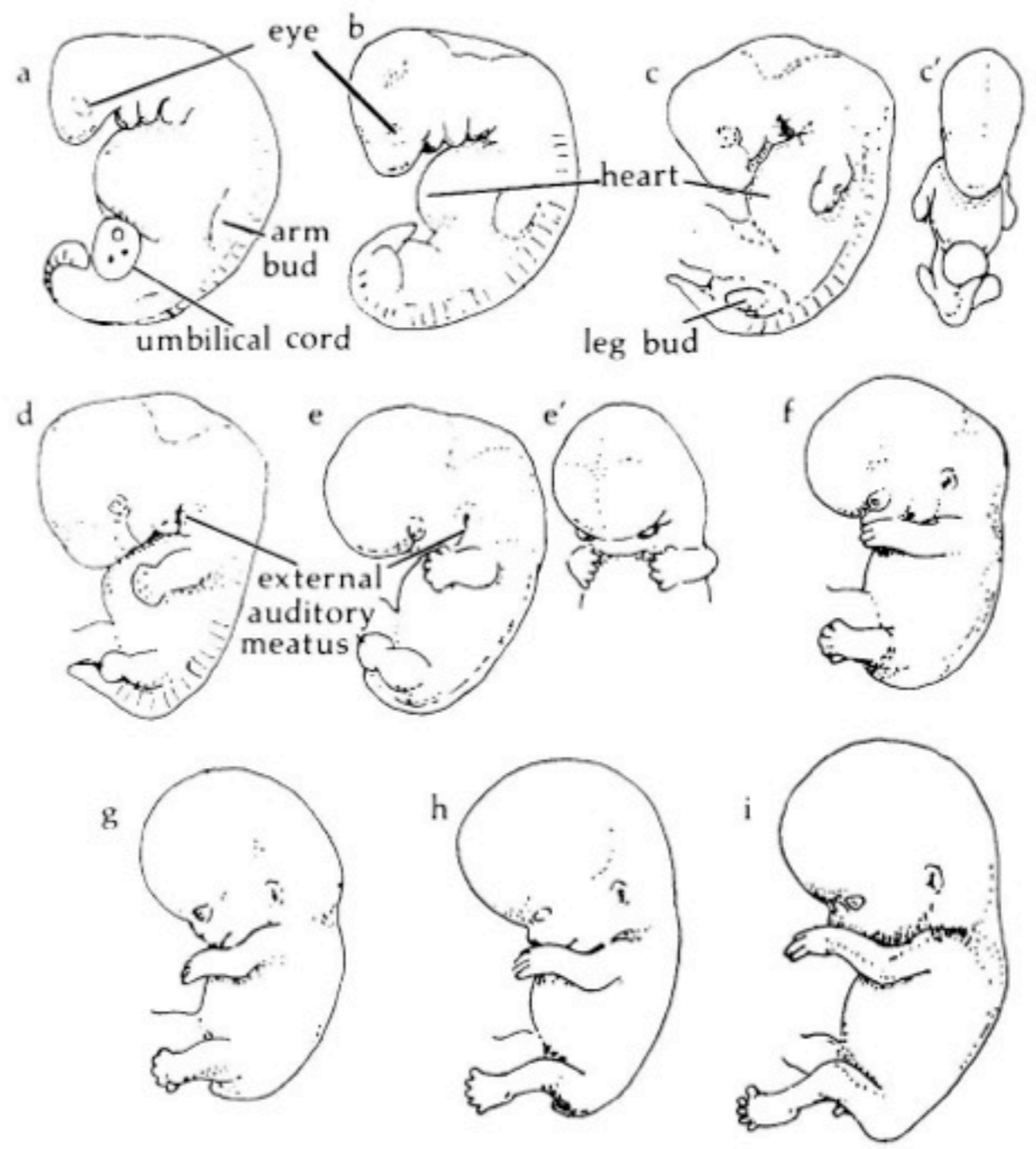
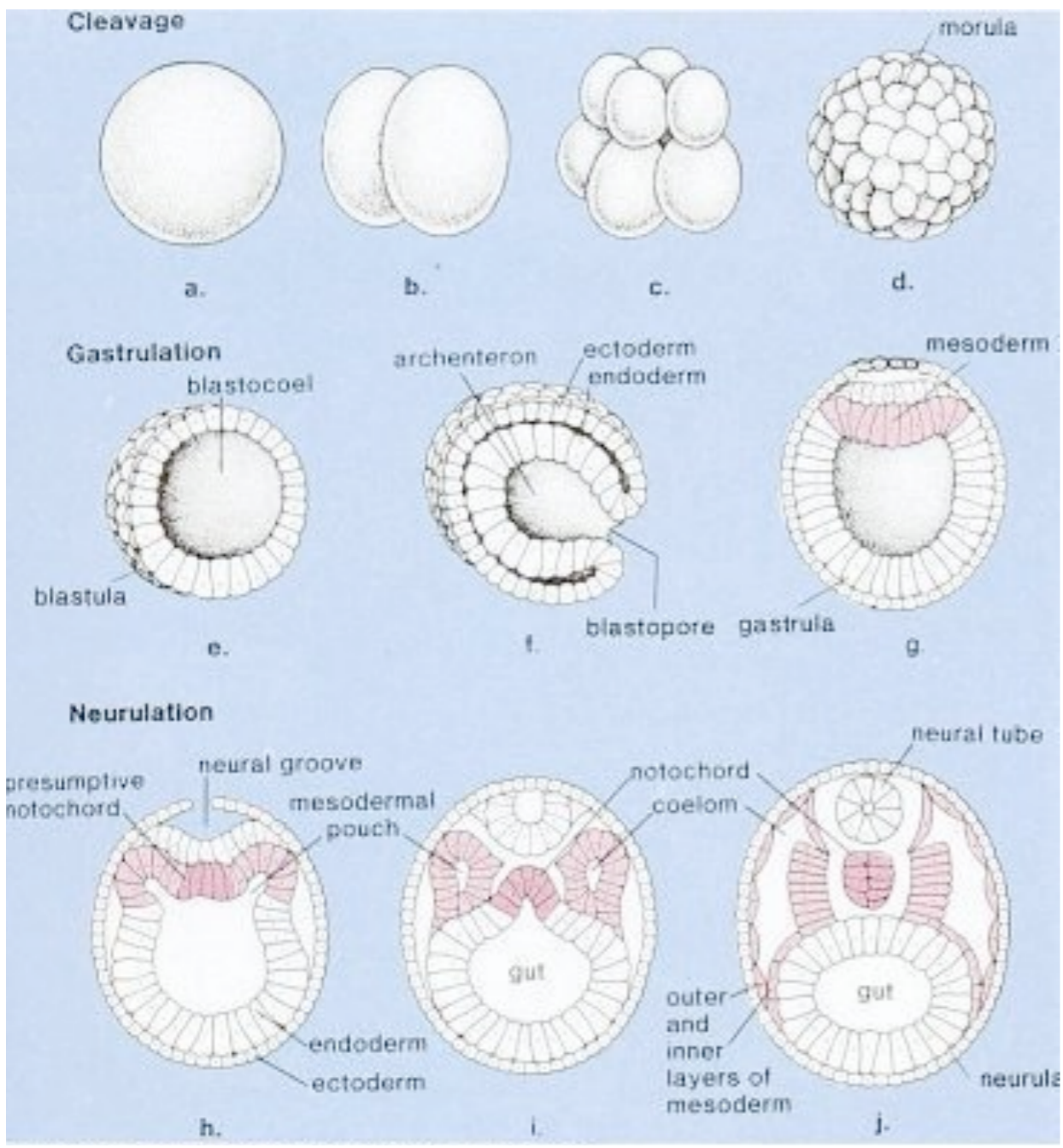
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Proteus Syndrome (AKT1) Overgrowth-Hypoglycemia (AKT2) / Hemimegalencephaly (AKT3)

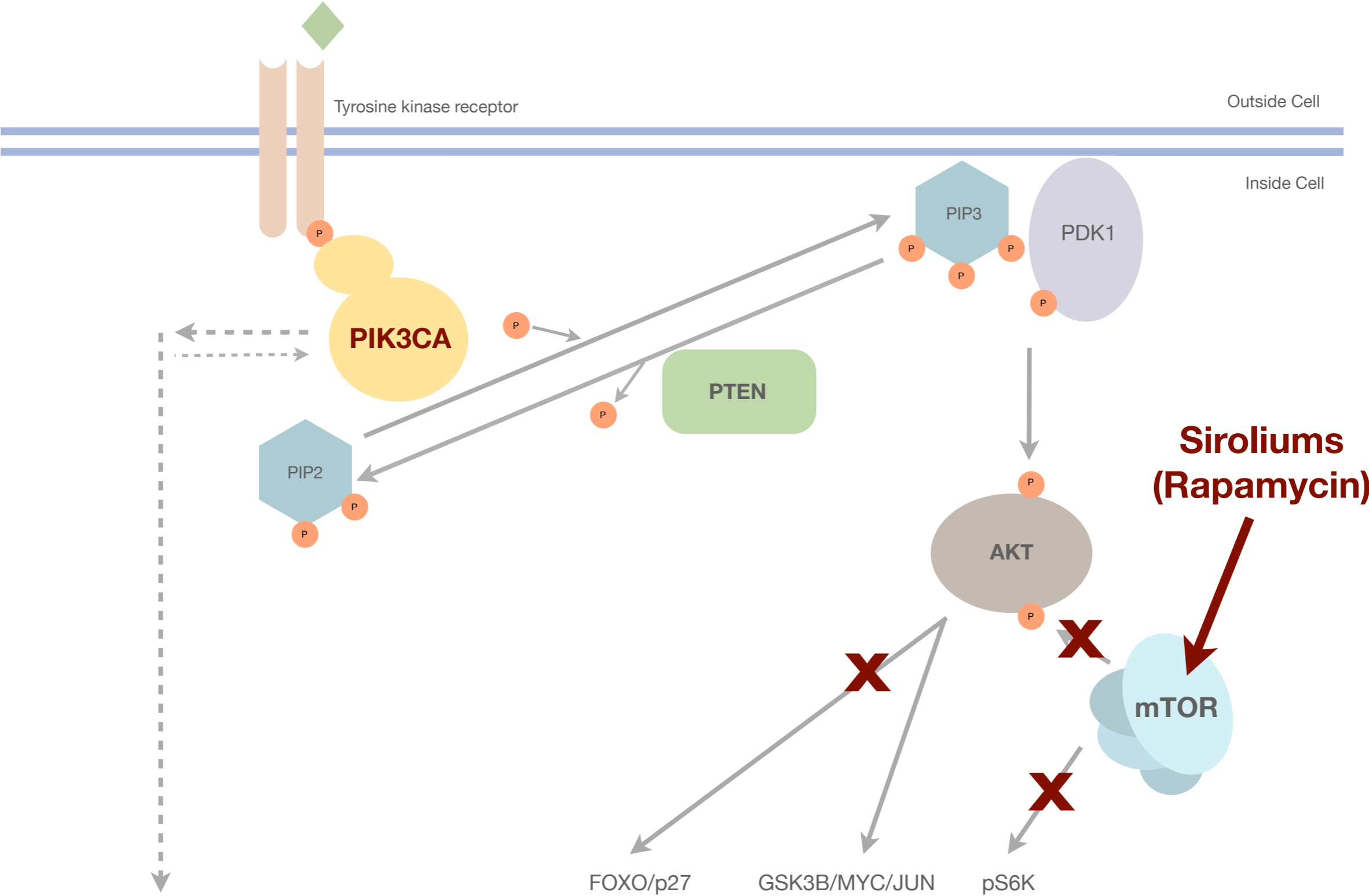


Cell growth, size, & proliferation regulation / fat metabolism

**CLOVES Syndrome/ Klippel-Trenaunay Syndrome (PIK3CA)
 Megalencephaly ± Capillary Malformation/ MPPH (PIK3CA, PIK3R2)**

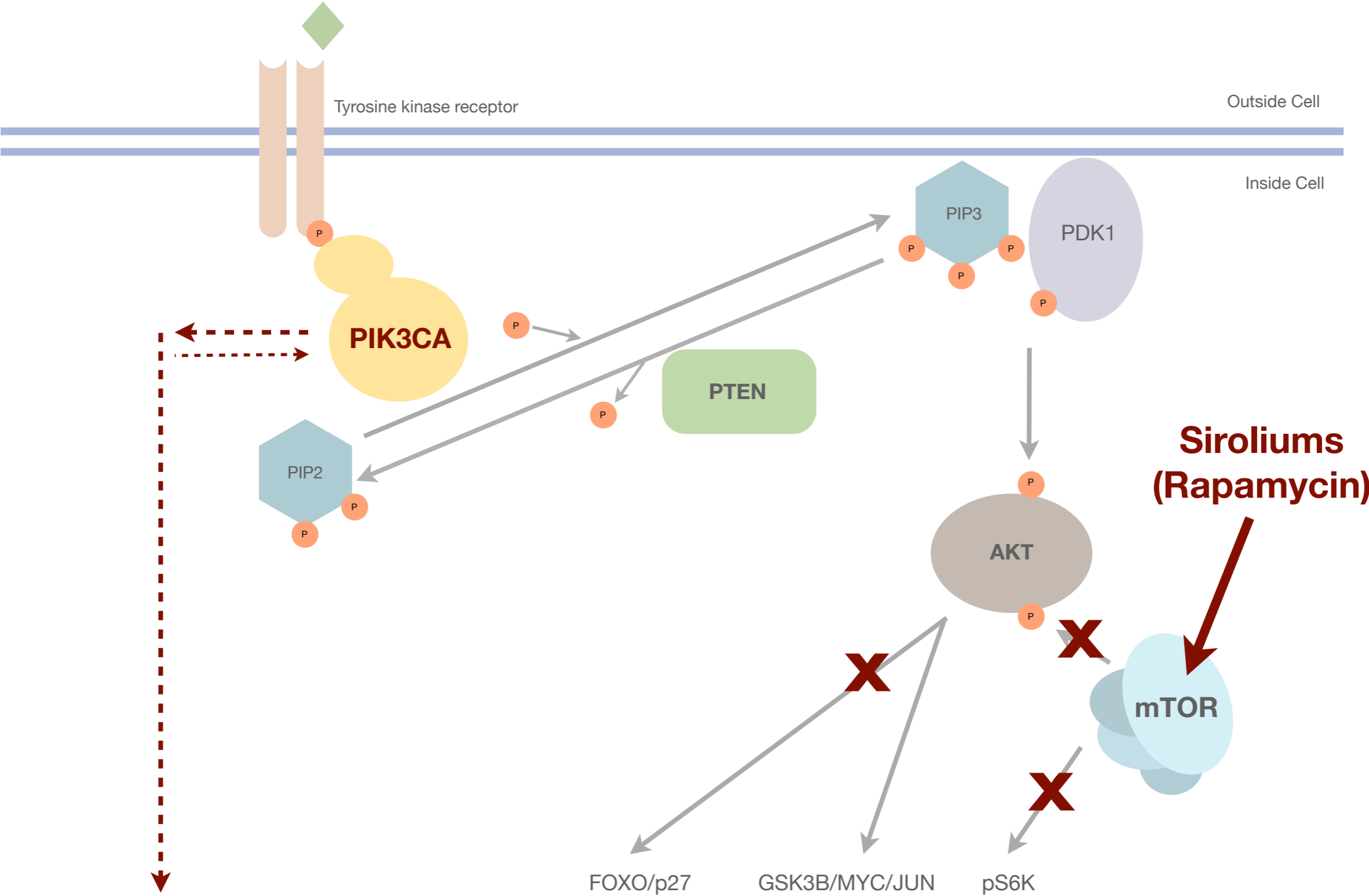


CLOVES Syndrome- New Therapies



Cell growth, size, & proliferation regulation / fat metabolism/ blood vessel growth

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Cell growth, size, & proliferation regulation / fat metabolism/ blood vessel growth

