# IVACAFTOR (KALYDECO®)

INFORMATION ABOUT THE DRUG AND GENETIC VARIATION

## What is ivacaftor?

- Ivacaftor is a medication that is used to treat cystic fibrosis in patients with specific mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene.
- Ivacaftor increases the ability of the CFTR protein to transport chloride.

## Genetic Variations

- A functional CFTR protein allows for normal chloride transport and more balanced water and salts. This results in mucus with normal thickness.
- A defective CFTR protein (caused by the G551D mutation in this case) causes decreased chloride transport, leading to an imbalance of water and salt absorption. This results in thick mucus.

## Drug-Gene Interaction

- The CFTR allele that ivacaftor treats is the rs75527207G>A variant, which causes the protein to be unable to transport chloride through the channel.
- Ivacaftor is strongly recommended for cystic fibrosis treatment in patients with one or two copies of the rs75527207G>A variant.

If you have any more questions please visit the website listed below or talk to a healthcare professional.

- http://drugsandgenes.com/
- http://www.pharmgkb.org