Presentation One Outline

* Opening assertion – Personal Story: At three days old, my little brother was diagnosed with a genetic disease called cystic fibrosis (picture)
  + AND this was extremely upsetting for my family, particularly my parents.
  + BUT it prompted them – and as I got older, me – to learn more about the disease and what this diagnosis meant for my brother
  + THEREFORE we will turn to a brief overview of CF
* Next assertion: Cystic fibrosis patients have a significantly shortened lifespan.
  + Source: CFF.org (Cystic Fibrosis Foundation – About CF)
  + In the 1950s, CF patients rarely lived long enough to attend, let alone graduate, from elementary school AND when my brother was born, prognoses were not much better. Treatments addressed symptoms, but not causes. BUT recent breakthroughs in treatment addressing causes of the disease have greatly increased lifespan THEREFORE patients are now living into their 40s and beyond.
* Next assertion: Cystic fibrosis can cause damage to the lungs, pancreas, and other organs due to a buildup of thick, sticky mucus.
  + Source: Cff.org
  + Other symptoms include salty skin; frequent, wet coughs; poor weight gain despite healthy appetite, and increased risk of infection (immunodeficiency)
  + This gives a basic overview of CF BUT the new treatments I’ll be discussing are so revolutionary because they address the root cause of CF rather than the symptoms, THEREFORE let’s examine the exact effect of the CF mutation
* Next assertion: The CF Delta F508 mutation has two main effects on operation of cells
  + Source: How Orkambi Works – from Orkambi website.
  + Include graphics on this slide!
  + My brother has a type of CF called Delta F508. This is the most common type of CF mutation, and also the one that the treatment we will discuss today addresses.
  + The F508del mutation has two main effects.
  + The first is that the body of an F508del CF patient is unable to produce as many normal CFTR proteins as a healthy body.
  + CFTR proteins transport chloride ions across a cell’s surface
  + Many of the mutated proteins are misshapen and ineffective, or else cannot make it all the way to the surface of the cell where they can be of use, or stay there as long.
  + The second effect is that the opening and closing of the channels created by the CFTR protein doesn't occur nearly often enough.
  + The result is a lack of chloride ions transported across the cell’s surface, and thus a buildup of mucus in the lungs and other areas.
  + This can lead to all kinds of other health problems.
* Final assertion: Orkambi is a miraculous breakthrough for patients with the Delta F508 CF mutation – addressing the cause, not just the symptoms
  + Source: How Orkambi Works
  + This drug is designed to be a partial to substantial cure for F508del CF patients, combining drugs called lumacaftor and ivacaftor.
  + Lumacaftor helps CFTR to form properly and remain on the cell’s surface for longer.
  + Ivacaftor helps improve the opening and closing of the chloride channel created by the CFTR protein.
  + This discovery is revolutionary for people living with cystic fibrosis, and has the possibility to substantially expand their expected lifespan over time.
  + THEREFORE, best of all, my brother was healthy enough to begin taking this drug almost immediately after its approval, a miracle for me and many others who love people with cystic fibrosis.

Bibliography

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