Should I Mortgage My House or Move to Another Country Doc?

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Introduction: Cystic fibrosis (CF) is a life-limiting genetic disease, resulting from abnormalities in the CF transmembrane conductance regulator (CFTR), a chloride channel found in cells of various organs. Over 2000 known disease-causing mutations that interrupt various stages of CFTR synthesis and function. Ivacaftor is a potentiator for the CFTR protein at the surface of epithelial cells, licensed and funded in the UK for patients above 2 years. Lumacaftor is a corrector, acts as a chaperone during protein folding and increases the number of CFTR proteins that are trafficked to the cell surface. Orkambi is a combination of Ivacaftor and Lumacaftor, licensed for patients DF508 homozygous, above 6 years old and funded in some countries, but not in the UK.

Case: 6.5 years old girl diagnosed with CF via new-born screening (DF508 homozygous). Stable from respiratory perspective. CF liver disease at the age of 5 years, on ursodeoxycholic acid. Progressive worsening of her liver USS imaging, synthetic function remains mostly normal. The family contacted a US CF centre to move her CF care and start Orkambi. They elected to purchase Orkambi privately in the UK. They could afford only a first 3 month prescription. After the 1st dose, she had a large bowel movement with fresh blood and continues to complain of abdominal discomfort. Stable clinical examination, baseline bloods/imaging.

Discussion points:

1. How should the medical CF team counsel families who would like to privately purchase Orkambi?

2. How should we monitor these patients?

- Is it right to divert NHS CF resources for the monitoring of patients to those families who can afford gene therapy?

3. Should physicians recommend patients a) move to countries where they will have free access to gene therapy? b) mortgage their house?

4. What do we think as professionals about parents driving treatment decisions?