**Title:** Successful off-label use of elexacaftor/tezacaftor/ivacaftor in an infant with severe CF disease

**Lay Title:** Early access to the CFTR modulator treatment helps severely ill infant with Cystic Fibrosis

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**What was your research question?**

Case report of elexacaftor/tezacaftor/ivacaftor in a young infant. Recently, many patients with Cystic Fibrosis (CF) have had access to powerful new medications called CFTR modulators. These drugs work by helping the faulty protein in CF function better. Different countries have different access based on their health systems.

**Why is this important?**

One of the most widely used CFTR modulators is a combination of three drugs elexacaftor/ tezacaftor/ivacaftor (ETI) with the trade name of “Trikaftor®”. Currently, ETI is only approved for older children and adults. Infants cannot access this potentially life-saving treatment through normal channels, even when they're desperately sick as the trials have not been completed yet.

**What did you do?**

This case discusses an infant girl with CF whose condition was far more severe than typical for her age. Her first year of life was dominated by hospital stays - seven admissions totalling 110 days in the hospital. Despite receiving a government-funded approved CFTR modulator medication (lumacaftor/ivacaftor) at 12 months old, her health continued to decline.

By 14 months, she had severe lung disease and ongoing significant nutritional problems. Her doctors had tried everything available to them for her age group, but nothing was working effectively.

**What did you find?**

Faced with this dire situation, the medical team - after extensive discussions involving multiple specialists, hospital administrators, and ethics experts – decided, with the consent of the parents, to provide ETI treatment "off-label" (meaning outside its approved age range). The health team estimated a dosage based on weight and the dose form that was currently available. The results were remarkable. After starting ETI:

* Her overall health improved significantly
* A key diagnostic test (sweat chloride) returned to normal levels
* She began gaining weight and growing properly
* She hasn't needed any hospital admissions since starting the treatment

**What does this mean and reasons for caution?**

This case demonstrates that ETI was helpful for this child with severe CF, however, one successful case isn't enough to change medical practice. This case emphasizes the critical need for formal clinical trials in infants to properly evaluate both the benefits and potential risks of ETI in this agegroup. This case also demonstrates the importance of conducting off label use very carefully with multidisciplinary support and consideration of ethical issues and informed consent. Clinical trials will provide the evidence needed to make this treatment routinely available to the youngest CF patients who might benefit from it.

**What’s next?**

This breakthrough offers hope for medical teams and families dealing with severe CF in very young children, while highlighting the importance of carefully conducted research to ensure new treatments are both safe and effective across all age groups.

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