Changing incidence of CF in Ireland?

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Ireland has the world’s highest incidence of Cystic Fibrosis at approximately 1 in 1400 [1,2]. CF was integrated into the newborn bloodspot screening programme in July 2011 using an IRT/DNA analysis strategy. Babies with a blood IRT above the 99th percentile and with one or two CFTR mutations detected are referred for sweat testing.

The anticipated number of CF cases (54) and carriers (94), were calculated based on the birth rates from the 2008 census and assuming a 2.5-fold enrichment of carriers within the top IRT percentile.

During the first 6 months of the programme, 37,435 babies were screened and 391 (1.0%) were referred for DNA testing. 16 CF cases (all with 2 mutations and confirmed by sweat testing) and 35 unaffected carriers were detected, giving an annualised CF incidence of 1 in 2340.

Just 59% of the predicted number of CF cases (p = 0.038) and 74% of the predicted number of unaffected carriers (p = 0.014) were detected. Data from 2010 indicates the number of unaffected carriers (p = 0.014) were detected, giving an annualised CF incidence of 1 in 2340.

Longitudinal changes in lung function and risk of death in cystic fibrosis: developing a joint model for the UK population

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Introduction: We outline a novel approach for the joint modelling of lung function and survival in the UK Cystic Fibrosis Population. The aim is to quantify how aspects of an individual cystic fibrosis patient’s longitudinal profile of %FEV1 are related to their survival prognosis.

Methods: We use the UK CF registry and apply recently developed methodology using open-source software (R) for the joint analysis of repeated measurements and time-to-event outcomes. These methods allow examination of association between %FEV1 and covariates such as sex, genotype and screening status, whilst allowing for correlation within patients, trends over time and potentially informative missing values. Key methodological challenges relate to accommodating cohort effects, and biased entry to registry cohorts.

Results: The dataset includes around 46,000 measures of %FEV1 on 8,000 patients seen between 1999 and 2010, and captures 1000 deaths. In our preliminary analysis stratified by birth cohort: For people born in 1975 to 1979, all other things being equal, a 10% higher level of %FEV1 is associated with a halving of the concurrent hazard for death (HR 0.44 CI 0.38 to 0.50). People with a stable %FEV1 compared to a 1% per year decline, have a 10% lower hazard of death for every year that passes (HR 0.9 CI 0.89 to 0.92).

Conclusions: We apply a novel modelling approach to quantify how longitudinal changes in lung function are related to survival. Both an increased rate of decline in lung function, and a decreased absolute level of lung function are associated with an increased risk of death.