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Comparative demographics of the European Cystic Fibrosis population: does EU membership confer an advantage?

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Background

Country-specific patient rare disease registries are rarely used to make international comparisons because of protocol discrepancies in data collation. Here, we attempt to overcome this limitation by using the inherited disease Cystic Fibrosis (CF) as a paradigm. CF provides a good example because its common form (homozygous F508del-CFTR) occurs across all European social strata appearing frequently but randomly thus providing an opportunity to measure health outcomes.

Methods

Country-specific CF Registries were combined cross-sectionally using a common data protocol (http://www.eurocarecf.eu) to compare patient demographics between the European Union (EU) and non-EU countries using EU membership in 2003 as a reference base. We tested the hypothesis that the nine-fold higher resources within the EU would translate into better outcomes.

Findings

Data were collected on age, age at diagnosis and CF genotype from 29,025 CF patients registered in 35 European countries. Median age was 16.3 years but was ~4.9 years older in EU countries (17.0 years) than non-EU countries (12.1 years; $p<0.001$; CI for the difference was 4-5.1 years, a significant difference (OR 2.4, 95% CI 1.9 - 3.0). Under-ascertainment was unlikely because the relative paucity of F508del-homozygous patients outside the EU was also significant (95% present clinically in childhood). We estimate that the current CF population of non-EU countries would rise by 84% if they had a CF demographic profile comparable to those of the EU countries who were already EU members in 2003.

Interpretation

Given that neither the CF carrier frequency nor the relative territorial population size is significantly different between the EU and non-EU participants, the reasons for this apparent deficit in CF patients of a common genotype in non-EU countries require explanation. It may be that under diagnosis and premature childhood mortality are the main drivers of the relative paucity of CF in non EU states.

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