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A Digital Health Approach: Using Electronic Health Records to Evaluate the Cost Benefit of Early Diagnosis of Alpha-1 Antitrypsin Deficiency in the UK

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Abstract : Alpha-1 antitrypsin deficiency (AATD) is a rare, genetic, and multisystemic condition. Underdiagnosis is common, leading to chronic pulmonary and hepatic complications, increased resource utilization, and additional costs to the healthcare system. Currently, there is limited evidence of the direct medical costs of AATD diagnosis in the UK. This study explores the economic impact of AATD patients during the 3 years before diagnosis and to identify the major cost drivers using primary and secondary care electronic health record (EHR) data. The 3 years before diagnosis time period was chosen based on the ability of our tool to identify patients earlier. The AATD algorithm was created using published disease criteria and applied to 148 known AATD patients' EHR found in a primary care database of 936,148 patients (413,674 Biobank and 501,188 in a single primary care locality). Among 148 patients, 9 patients were flagged earlier by the tool and, on average, could save 3 (1-6) years per patient. We analysed 101 of the 148 AATD patients' primary care journey and 20 patients' Hospital Episode Statistics (HES) data, all of whom had at least 3 years of clinical history in their records before diagnosis. The codes related to laboratory tests, clinical visits, referrals, hospitalization days, day case, and inpatient admissions attributable to AATD were examined in this 3-year period before diagnosis. The average cost per patient was calculated, and the direct medical costs were modelled based on the mean prevalence of 100 AATD patients in a 500,000 population. A deterministic sensitivity analysis (DSA) of 20% was performed to determine the major cost drivers. Cost data was obtained from the NHS National tariff 2020/21, National Schedule of NHS Costs 2018/19, PSSRU 2018/19, and private care tariff. The total direct medical cost of one hundred AATD patients three years before diagnosis in primary and secondary care in the UK was £3,556,489, with an average direct cost per patient of £35,565. A vast majority of this total direct cost (95%) was associated with inpatient admissions (£3,378,229). The DSA determined that the costs associated with tier-2 laboratory tests and inpatient admissions were the greatest contributors to direct costs in primary and secondary care, respectively. This retrospective study shows the role of EHRs in calculating direct medical costs and the potential benefit of new technologies for the early identification of patients with AATD to reduce the economic burden in primary and secondary care in the UK.

Keywords : alpha-1 antitrypsin deficiency, costs, digital health, early diagnosis
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