



Background

- Fragile X Syndrome (FXS) is a genetic condition that is associated with cognitive impairment, a distinct physical phenotype and intellectual disability (1,2).
- At present, treatment generally aims to manage commonly co-occurring physical and mental health conditions e.g., epilepsy, anxiety and ADHD.
- The rationale behind this study centres around a lack of data as to the level of resource use and costs associated with treating Fragile X within the UK.

Methods

- Patients were selected from the Clinical Practice Research Datalink (CPRD) Aurum dataset linked to Hospital Episode Statistics (HES).
- CPRD Aurum is derived from primary care practices in England and captures approximately 20% of the England population.
- Patients with FXS were selected by either medcodes (Aurum) or ICD-10 codes (HES) (Table 1).
- FXS patients were required to be registered at an Aurum practice and have a diagnosis before 1st January 2019.
- Patients with FXS were matched 1:1 to non-FXS control patients on age, gender and concurrent practice registration.
- Primary care contacts and costs, including prescriptions, were taken from CPRD Aurum datasets. Inpatient, outpatient and emergency department contacts and costs were taken from HES.
- All healthcare categories were costed according to the relevant consultation and associated tariff (3, 4, 5).
- Healthcare contacts and associated costs (UK 2019/2020 prices) were calculated per person year (PPY) for the year 2019.
- Generalised linear models were constructed to compare the incidence rate ratio (IRR) (Poisson) and cost ratio (CR) (Gamma) between patients with FXS and controls.

Table 1. The clinical codes used to select patients with FXS.

Description	Clinical code	Type
Fragile X Syndrome	2090010, 893501000006110, 940371000006118	Medcode
Fragile X Chromosome	315486012	Medcode
Cause of learning disability: Fragile X Syndrome	1009571000006115	Medcode
FRAXA – Fragile X Syndrome	2508311000006110	Medcode
Martin-Bell Syndrome	2508291000006111	Medcode
Fragile X Chromosome	Q99.2	ICD-10

Results

- 3,040 patients were included in the analysis (n=1,520 cases and controls)
- The mean age was 32 years and 65% were male.
- Patients with FXS had a higher number of contacts in primary care, inpatient, outpatient and Emergency Department (ED).

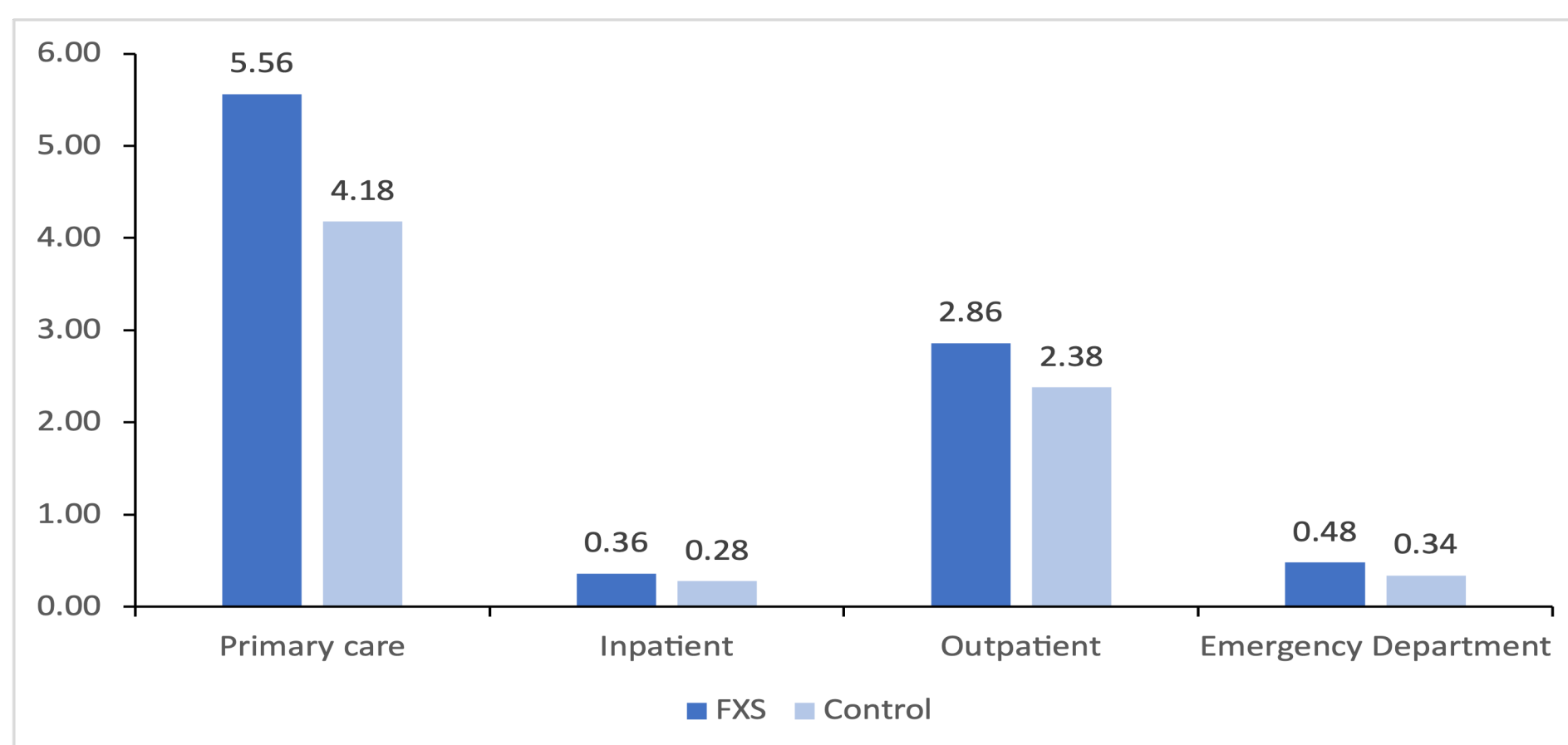


Figure 1. The rate (PPY) of healthcare contacts for patients with Fragile X and matched controls from the Clinical Practice Research Datalink and Hospital Episode Statistics 2019.

- The rate of healthcare contacts PPY was significantly greater for FXS patients than controls in all healthcare settings.

	IRR (95% CI)	p-value
Primary care	1.19 (1.15–1.23)	<0.0001
Inpatient	1.18 (1.02–1.37)	0.0293
Outpatient	1.11 (1.05–1.16)	<0.0001
Emergency Department	1.28 (1.12–1.48)	0.0005

Figure 2. Generalised linear model for healthcare contacts compared between patients with Fragile X and matched controls.

- Patients with FXS also had higher costs in each healthcare setting than controls.

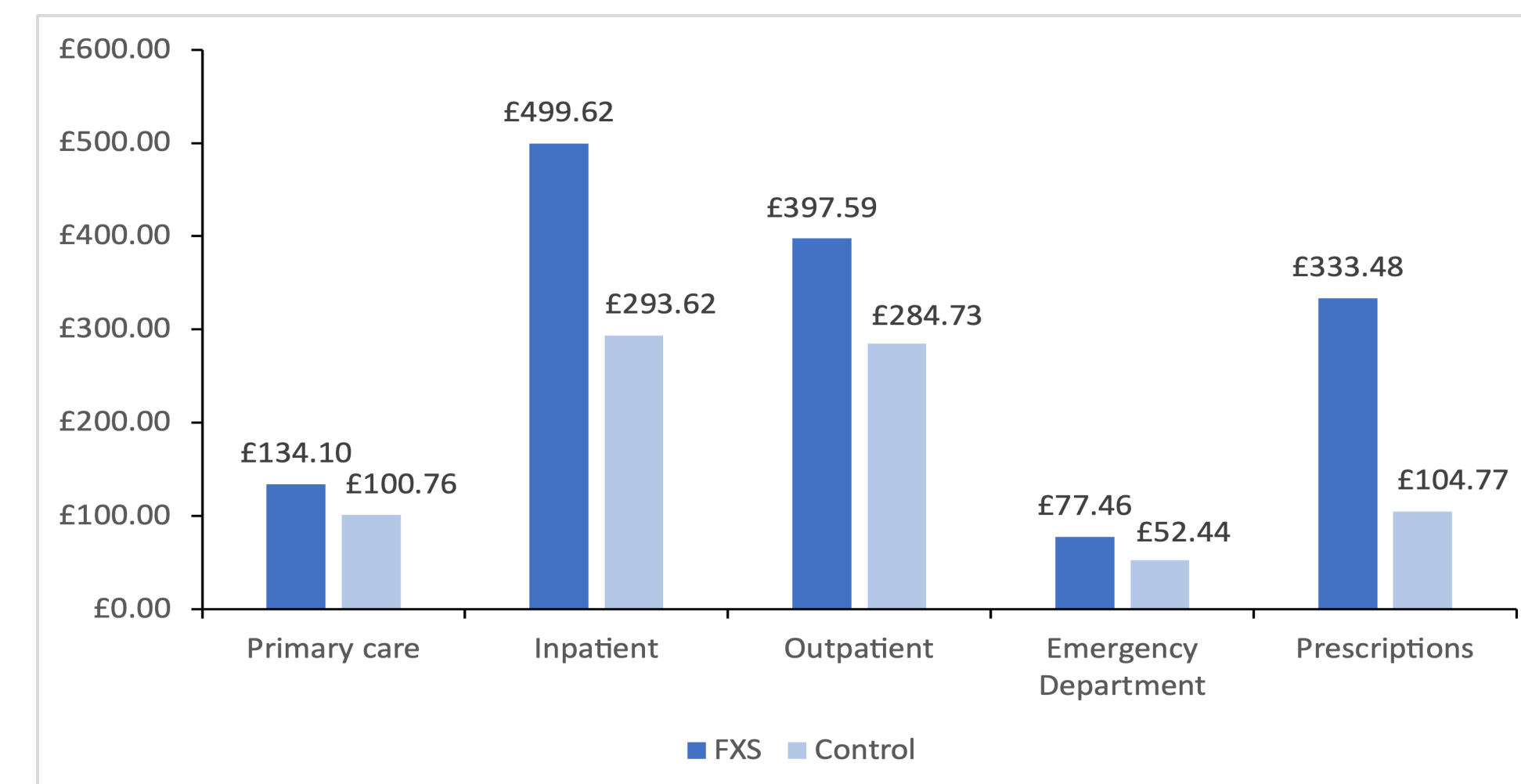


Figure 3. The healthcare cost (PPY) for patients with Fragile X and matched controls from the Clinical Practice Research Datalink and Hospital Episode Statistics 2019.

- Costs PPY were also significantly greater for cases than controls for primary care, inpatient, outpatient and primary care prescriptions.
- There was no significant difference in ED costs between cases and controls.

	CRR (95% CI)	p-value
Primary care	1.14 (1.03–1.27)	0.0130
Inpatient	1.67 (1.19–2.35)	0.0031
Outpatient	1.29 (1.05–1.58)	0.0139
Emergency Department	1.08 (0.97–1.21)	0.1767
Prescriptions	2.69 (2.09–3.47)	<0.0001

Figure 4. Generalised linear model for healthcare costs compared between patients with Fragile X and matched controls.

Conclusion

- The findings from this study identified that FXS patients had a significantly higher rate of contacts per person year than control patients in primary care, inpatient, outpatient and ED.
- For healthcare resource use costs, FXS patients in the prevalent population had significantly higher costs per person year than control patients in primary care, inpatient, outpatient and primary care prescriptions.
- A limitation of the study is that prescriptions issued in secondary care are not recorded in CPRD, and therefore the cost of prescriptions in the population may be underestimated.

References

1) Fragile X Society. About Fragile X syndrome [Internet]. 2021. Available from: <https://www.fragilex.org.uk/syndrome>. 2) National Fragile X Foundation. Fragile X 101 [Internet]. Available from: <https://fragilex.org/understanding-fragile-x/fragile-x-101/>. 3) Personal Social Services Research Unit. Unit costs of health and social care [Internet]. 2020. Available from: <https://www.pssru.ac.uk/project-pages/unit-costs/>. 4) NHS Business Services Authority. Prescription cost analysis - England 2019 [Internet]. 2020. Available from: <https://www.nhsbsa.nhs.uk/statistical-collections/prescription-cost-analysis-england/prescription-cost-analysis-england-2019>. 5) NHS England. National tariff 2019/20: Documents and policies [Internet]. 2020. Available from: <https://www.england.nhs.uk/publication/past-national-tariffs-documents-and-policies/>