Abstract Number: 5PSQ-015

# When seizures persist: exploring novel therapeutic options in Aicardi syndrome – case report

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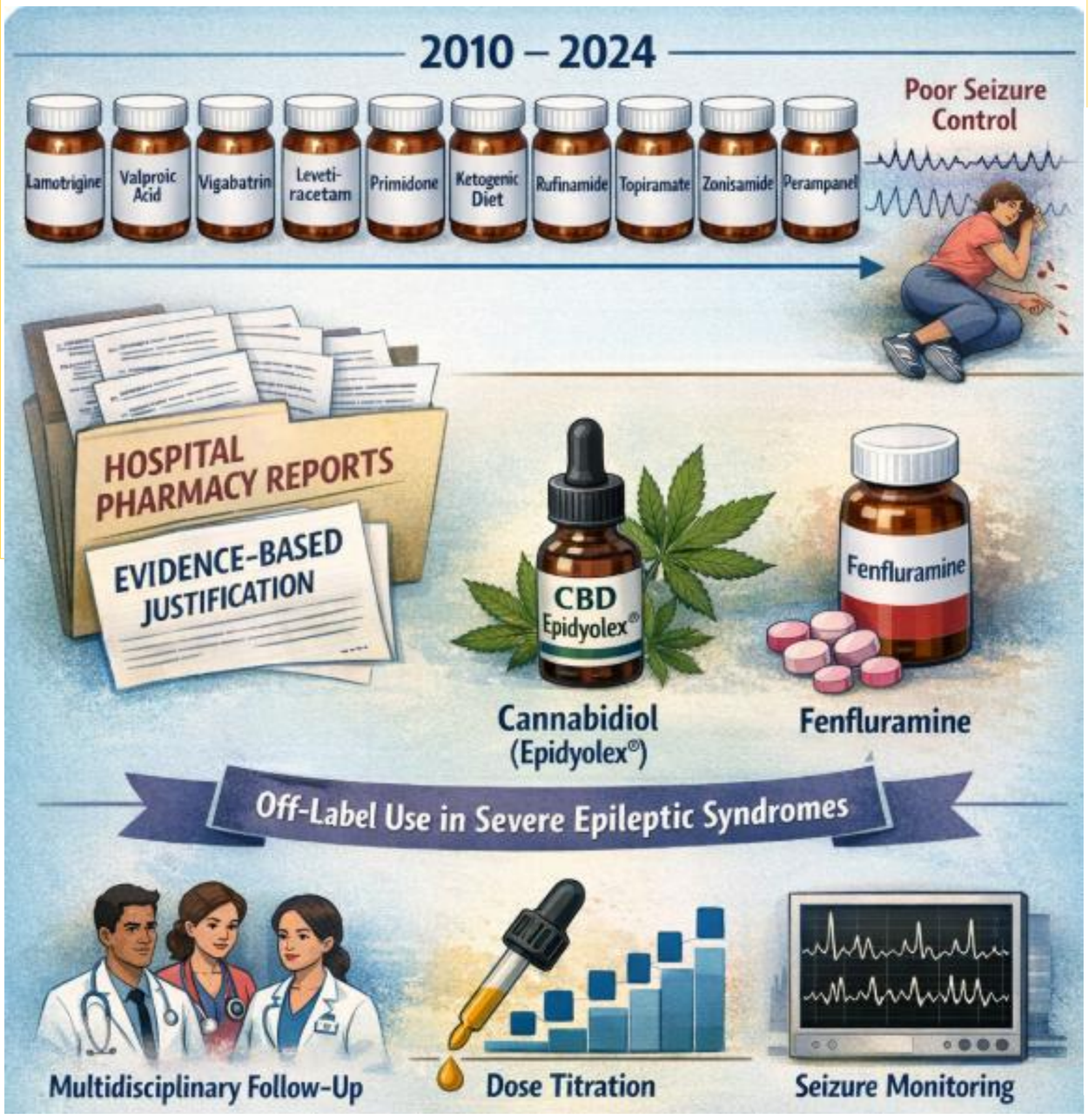
## BACKGROUND AND IMPORTANCE

Aicardi syndrome is a rare neurodevelopmental disorder characterised by agenesis of the corpus callosum, chorioretinal lacunae, and seizures. No treatments are approved specifically for this condition, and seizure control is often difficult. Drug-resistant epilepsy carries high morbidity and mortality, with few effective interventions once standard therapies fail.

## AIM AND OBJECTIVES

To describe the clinical evolution of a paediatric patient with Aicardi syndrome and drug-resistant epilepsy, and the impact of cannabidiol and fenfluramine treatment supported by hospital pharmacy intervention. The patient, a female diagnosed at 2 months of age, presented with frequent epileptic seizures unresponsive to multiple anti-seizure medications and dietary therapies.

## MATERIAL AND METHODS




## RESULTS

### Treatment Timeline & Clinical Outcomes

Cannabidiol · Perampanel · Fenfluramine · Cenobamate

**2022**

**Cannabidiol (CBD)** — DEC 2022 – INITIATION
Marked reduction in tonic–clonic seizures at initiation. Sustained partial benefit maintained over 2-year follow-up.
TC seizures ↓↓

**2023**

**Reflex Seizures** — 2023 – DOSE OPTIMISATION
Persistent reflex seizures despite TC control prompted CBD dose optimisation and withdrawal of perampanel.
Partial response

**Perampanel** — 2023 – WITHDRAWN
Discontinued as part of dose optimisation strategy due to persistent reflex seizure activity under combined therapy.
Withdrawn

**2024**

**Fenfluramine** — 2024 – 10-MONTH TRIAL
Introduced as add-on therapy; discontinued after 10 months due to lack of clinical benefit. No severe adverse effects.
No benefit

**Cenobamate** — 2024 – TRIALLED
Trialled as alternative antiseizure medication. No improvement in seizure control observed.
No improvement

| 2 yr | 4 | 2 | 0 |
|---|---|---|---|
| SUSTAINED CBD BENEFIT | MEDICATIONS TRIALLED | DISCONTINUED (NO BENEFIT) | SEVERE ADVERSE EVENTS |

✓ **Safety profile:** No severe adverse effects were observed with either cannabidiol or fenfluramine throughout the entire treatment period.

## CONCLUSION AND RELEVANCE

Cannabidiol provided substantial early seizure reduction with sustained partial benefit over more than 2.5 years of continuous treatment — a follow-up period exceeding that reported in the published literature for Aicardi syndrome and most other epileptic encephalopathies. This long-term observation highlights both the potential durability of cannabidiol's effect and the need for systematic studies beyond 48 weeks. Fenfluramine use in Aicardi syndrome, with no robust evidence base, was ineffective in our patient. Similarly, cenobamate lacks supporting data in this population and did not improve seizure control. This case underlines the critical role of hospital pharmacy in bridging evidence gaps, enabling access to novel therapies, and ensuring rigorous monitoring in highly refractory contexts. Continuous reassessment and long-term data collection are essential to advance the therapeutic landscape for rare epilepsies.