

MEDICINAL CANNABIS USE IN RARE DISEASES

Jekaterina Parovincaka, Janis Vella Szijj, Anthony Serracino Inglott

Department of Pharmacy, Faculty of Medicine and Surgery, University of Malta, Msida, Malta

Email: janis.vella@um.edu.mt

INTRODUCTION

Rare diseases (RDs) affect over 300 million people worldwide. There are approximately 7,000 recognised RDs¹. RDs are usually genetic and are associated with severe debilitating symptoms which persist for a patient's lifetime². RD patients face multiple challenges including delay in accurate diagnosis and significant unmet need for beneficial therapies. For 90 % of RDs there are no effective therapies³. Medicinal Cannabis (MC) is used to relieve symptoms, such as pain, muscle spasticity and anxiety, which may be commonly experienced by patients with RDs. Being able to manage the symptoms and the progression of diseases, can positively impact patients, families and health care systems⁴.

AIM

To identify RDs for which MC is of interest.

METHOD

A systematic literature review was carried out. Open access peer review journal articles, published in English in PubMed Central or MEDLINE databases between January 2011 – September 2021 were included.

RESULTS

- ❖ 239 articles were identified, 36 articles included in the review
- ❖ 24 out of 36 articles included publications published in years 2020 and 2021
- MC was used in **22 different RDs**
- **20 of 22 RDs** have only symptomatic treatment available
- **15 of 22** Rare Diseases are of childhood onset

Medicinal Cannabis was mostly used in the treatment of:

Refractory seizures (n=12)

Pain / muscle spasticity (n=6)

- The most often used cannabinoid was **CBD**

Table 1: Cannabinoids used in patients with RDs

Cannabinoids used	Number of articles
CBD (Cannabidiol)	19
Cannabis-based medicines (CBMs)	8
Synthetic cannabinoids	5
THC: CBD (in 1:1 ratio)	2
CBDA (cannabidiolic acid)	1
Non-psychoactive doses of Medicinal Cannabis	1

Medicinal Cannabis use in Rare Diseases

Systemic diseases (n=8)	Systemic sclerosis
	Cystic fibrosis
	Tuberous Sclerosis Complex (TSC)
	Scleroderma (Systemic sclerosis)
	Systemic lupus erythematosus
	CDKL5 Deficiency Disorder
	Dermatomyositis
	Neurofibromatosis Type 1
Epileptic conditions (n=6)	Dravet syndrome
	Lennox-Gastaut Syndrome
	Febrile infection-related epilepsy syndrome (FIRES)
	Myoclonic atonic epilepsy (Doose syndrome)
	Infantile spasms (West syndrome)
Neurodegenerative and neurological diseases (n=4)	Lafora Disease
	Amyotrophic Lateral Sclerosis
	Spinocerebellar ataxia type-3
	Huntington's Disease (Juvenile and Pediatric manifestation)
Neurodevelopmental disorders (2)	Multiple sclerosis
	Rett syndrome
Dermatological condition (1)	Williams-Beuren Syndrome
Gastrointestinal disorder (1)	Epidermolysis bullosa
	Chronic Intestinal Pseudo-Obstruction

CONCLUSION

Literature suggests that Medicinal Cannabis can be used in certain Rare Diseases.

In lack of efficacious treatment options, Medicinal Cannabis can be an alternative therapy for symptom relief, such as: seizures, pain and muscle spasticity.

REFERENCES

1. Nguengang Wakap S, Lambert DM, Olry A, Rodwell C, Gueydan C, Lanneau V, et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet. 2020;28(2):165-73.
2. Orphanet. About rare disease. [Cited on 30 January 2022]. Can be accessed from URL: http://www.orpha.net/consor/cgibin/Education_AboutRareDiseases.php?lng=EN
3. Groft SC, Posada M, Taruscio D. Progress, challenges and global approaches to rare diseases. Acta Paediatrica. 2021;110(10):2711-6.
4. Zanello G, Chan CH, Pearce DA. Recommendations from the IRDiRC Working Group on methodologies to assess the impact of diagnoses and therapies on rare disease patients. Orphanet Journal of Rare Diseases. 2022;17(1):1-0.