

# Dopa-responsive dystonia/Early-onset Parkinsonism

## Abstract

A patient's experience of living with Dopa-responsive Dystonia/Early-onset Parkinsonism is outlined. The way contacts with healthcare professionals could have been better are described. The medicines that are being taken, the elements of service provision that have been found to be most helpful and the steps needed to improve the ongoing management of the condition are identified. Key messages for healthcare professionals that have arisen from the patient experience are indicated.

**Keywords:** medical condition, medicines, pain, spasms

## About your medical condition

### What is the medical condition that is most important to you and is being presented here?

Dopa-responsive dystonia/Early-onset Parkinsonism.

### Can you please explain the problems you experience with this medical condition?

My problems began with pain and posturing of my left foot and leg. Over time this has progressed to muscle spasms in both feet, legs and trunk – it can be almost everywhere throughout my body. My body becomes fixed and I experience extreme pain. This can happen at any time of the day or night. The pain worsens with movement although I generally have more attacks at night, which causes me to wake up frequently. Daily, I am very tired and very slow. I usually feel better in the mornings than the afternoons - evening times I am extremely tired and unable to function so well. I walk around the house and can take a few steps with crutches. Out of the house I use a wheelchair. More recently, I am unable to sit up for any length of time.

### Can you please say how the medical condition was first diagnosed?

At age eleven I started attending high school. I became much more active and played lots of sports. My left leg and foot became much more painful and twisted. I began to have major problems with participating with sports and eventually even walking. The problems were investigated. I saw many types of doctors of varying specialities. Eventually, I was seen by a neurologist and admitted to a specialist hospital. After many tests and at age 17 years, I was diagnosed with Dystonia. Later, I was seen by a movement disorder specialist doctor and was further diagnosed with Early-onset Parkinsonism.

### Can you please say when the medical condition was first diagnosed?

1985.

### If you look back, what would you have liked to have been different in terms of contact with health professionals, etc?

- Faster diagnosis.
- Greater understanding of the condition by healthcare professionals.

## About your medicines

### Please list the medicines you taking for your medical condition.

- Lansoprazole 30mg gastro-resistant capsules  
Take one every day
- Bisacodyl 5mg gastro-resistant tablets  
Take one to two at night
- Miragbegron 50mg modified-release tablets  
Take one every day
- Nitrofurantoin 50mg tablets  
Take one at night
- Gabapentin 300mg capsules  
Three to be taken four times a day
- Tramadol 50mg capsules  
Take two every 4-6 hours
- Domperidone 10mg tablets  
Take two three times a day

- Celecoxib 200mg capsules  
One or two to be taken every day
- Sinemet CR 50mg/200mg tablets  
One to be taken five times a day
- Baclofen 10mg tablets  
Take 3 in the morning, 2 at lunch time, 2 at tea time and 3 at bed time
- Dantrolene 25mg capsules  
Take two tablets three times daily
- Lorazepam 1mg tablets  
Take half a tablet when required

Medication is clearly a time consuming, ever changing, constant in my life. I have a wonderful partner who keeps us constantly on top of things; not least in having to collect prescriptions on a constant basis!

We have a wonderful GP, with electronic communications for all prescription needs. Any queries are dealt with the same day with a telephone consultation. New and repeat requests are sent electronically from the GP to the pharmacy, and are ready for collection in good time.

Each day my medication is set out in a '1 Week Pill Chest', which has seven removable containers - one for each day. Each is split in to four sections: breakfast, lunchtime, teatime and bedtime. It is not filled by the pharmacy - it is done here at home. Most of the tablets are divided in to breakfast, lunch, dinner and bedtime sections. For the other, additional, tablets I have a reminder set on my phone.

Most of the time I don't really require any additional help as my body 'knows' when it is medication time. This of course falls apart if I am unwell - when this happens I always have someone around to help me take the correct medication at the correct times.

### Have you had any particularly bad experiences with regard to your medicines? If so, please explain and indicate how this could be avoided in future.

- Tiredness  
This can be overcome by pacing myself throughout the day e.g. I wake up early at 6.00am to take medication. I then need to have an hour or so resting whilst the medication begins to work and the side effects wear off.
- Constipation and sickness  
Although these may seem like routine everyday problems, they have a major effect on my day to day life. These are managed by my GP.
- Assumptions by healthcare professionals that new problems are side effects of recent medication changes when in fact they are due to new un-related conditions. I have been prescribed new medication by hospital consultants but, when home and taking the new medication, I have suffered sickness, headaches, and diarrhoea. These have been attributed to the new medication's side effects by healthcare professionals when the new symptoms were actually due to a bug contracted whilst in hospital, or a urinary tract infection (UTI), and nothing to do with the side effects of the new medication. This could be overcome by the GP having greater awareness of any unusual medication that is prescribed, doctors being mindful that other 'routine' illnesses can occur alongside a long-standing and more complicated condition and being under the direct supervision of the hospital consultant until the new medication is settled.

### Have you had any particularly good experiences with regard to your medicines? If so, please explain.

- Sinemet tablets made a huge improvement to my day-to-day life. I experienced less pain and much fewer spasms. I would say that this drug has changed my life.
- Nitrofurantoin has reduced the number and frequency of UTIs. This is a significant improvement.
- Pain medication is always helpful.

## About the services you received

### What have you found to be most helpful to you in terms of the services you have received?

All of the services I have received have been helpful. I am very thankful for the care that I receive. Most helpful have been:

- specialist knowledge and diagnosis of a Consultant Neurologist and a Movement Disorder Specialist Professor.
- continued and constant help, support and understanding by the GP Practice.

- a prompt and accurate service from the local pharmacy. Often, because of contact with multiple doctors and departments, drugs may be issued that are contraindicated. The Pharmacist is often the most important person to notice this and avoid problems arising.
- the provision of an electric wheelchair by Wheelchair Services has been helpful in allowing me to leave the house in a safe and comfortable way.

## To what extent have the health professionals you have come in contact with appreciated what it was like from your position as a patient?

This has been varied and patchy. In specialist hospitals and departments, consultant knowledge and awareness has been good. When in contact with other healthcare professionals, however, understanding has been poor, such as:

- nurses and healthcare assistants have NO understanding
- physiotherapists have very little understanding
- GP understanding is varied. One GP said that he knew nothing about the condition and told me to look it up on the internet. Another GP told me they have no understanding of the condition and I need to discuss everything (even something as routine as a cold) with my specialist consultant. They did not appreciate what it is like for me at all. Other GPs have a good understanding and try to appreciate what it is like from my position.

## To what extent was the information you were given about your medical condition sufficient for you?

At first, the information I was given was poor. At the time of diagnosis I was told how to spell the condition, but nothing about what it would mean to me. I was directed to The Dystonia Society for further information. Later, specialist consultants have provided me with good information.

## What have been the best experiences you have had with the services you have received?

- Specialist Consultants.
- Care given by GPs after such poor experiences with earlier GP Practices.
- Pharmacy services.

## About other medical conditions

Do you have just one medical condition that make life problematic for you? If 'No', please list the other medical conditions and explain the main problems you experience with each one:

Medical condition	Main problem experienced
Oesophageal spasm	Thoracic pain
T5 spinal cord hernia due to arachnoid cyst	Thoracic pain
Transverse myelitis	Weakness and numbness in lower limbs
Degenerative spine disease	Thoracic and Lumbar pain
Neuro-inflammatory disease	Thoracic pain

## About going forward

### What would you like to happen at this stage that would make living with your condition easier for you?

- Pain reduction
- A reduction in the number and severity of spasms
- Be able to walk around outside
- Reduced tiredness.

All of these I would like to happen but I realise it is unlikely that they will be achievable - but you did ask the question!

### If you could give a brief message to healthcare professionals, what would it be?

Thank you for taking care of me.

### Please add any other comments or observations that would be helpful to health professionals who are responsible for providing services for you.

Please do not see me simply as 'a patient with a disease or condition that you are treating' but as a complete person with many other conditions that interact with each other.

Also, see me as a person - not simply as a medical condition.

### What are the three most important things that health professionals should learn from your experiences?

- 1) Raise awareness of Dopa-responsive dystonia and other Early-onset Parkinsonism conditions.
- 2) Patients have other conditions as well as the one they are treating.
- 3) I am the same person I was before I needed to use a wheelchair - I can still think and talk for myself.

## Declaration of interests

**In the spirit of being open and transparent, would you please disclose any payments, interests or activities that could be perceived as influencing what you have written.**

A fee was offered by Pharmacy Management to complete this questionnaire within a defined timescale. I have no other interests to declare.

### KEY LEARNING POINTS FOR HEALTH PROFESSIONALS IDENTIFIED AT THE EDITING/PEER REVIEW STAGES

- Having lived with a debilitating long-term condition, we should remember that some patients are experts in monitoring and managing their condition. The implication of this is that we should ask the patient what works or does not work before offering advice.
- An important part of living with a debilitating, long-term condition is the burden of appointments and visits to pharmacies to collect medicines. The attendance at clinic appointments or at pharmacies for the collection of prescriptions or advice should be supported and made as easy as possible e.g. scheduling of supply of all medicines at the same time, better communication between primary and secondary care. Patients who do not have another person in the household to help them manage their medicines may be particularly at risk of drug-related misadventure.
- If we are privileged enough to have a patient with a rare, debilitating long-term conditions present to us, we should start by getting to know the person rather than the patient with a disease and a list of medicines. We should then read up on and learn about their disease in a way that leads to the patient receiving better drug related advice from us.