NPTR VIDEO CATALOG ENTRY

Disease	Pompe disease. Glycogen storage disease (GSD) type II. Carbohydrate disorders.
Title	Late-onset Pompe disease - An 18-Month Diagnostic Odyssey.
Link	https://www.youtube.com/watch?v=bTcVRd4najY
Key words	Pompe disease. Glycogen storage disease type II. Carbohydrate disorders. Patient/family experience. Symptoms. Genetics/genetic testing. Diet/treatment.
Description	In this video, the patient walks the viewer through her journey to find out her diagnosis of Pompe disease. She describes her symptoms, some of her diagnostic tests, and her treatment plan; she concludes by noting the impact of her treatment.
Length (min:sec)	3:28
Speaker(s) Background	Patient.
Objectives	1. List some symptoms of late-onset Pompe disease and the course of the disease. 2. Identify reasons why these symptoms might be overlooked. 3. Describe the potential benefits from enzyme replacement therapy.
Educational utility	Provides a personal narrative about living with the disease but should be accompanied by significant medical teaching to increase understanding about the disorder.
Technical aspects	Excellent audio and good video quality.
Relevant/ Target Audience	Health professionals and specialists. Trainees and students in the health professions. Other stakeholders who have an interest in the inborn errors.

Promotional aspects	This video discusses enzyme replacement therapy but does not promote a specific commercial product, treatment, and/or medical device.
Source	Rare Disease Report.
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