sNPTR VIDEO CATALOG ENTRY

Disease	Tyrosinemia type I. Hepatorenal tyrosinemia. Amino acid disorders.
Title	Tyrosinemia: Ultra-Rare, Life-Threatening, and Treatable.
Link	https://www.youtube.com/watch?v=omBQe5K-qHw
Key words	Tyrosinemia type I. Hepatorenal tyrosinemia. Amino acid disorders. Patient/family experience. General disease overview. Symptoms. Diagnostic testing. Biochemistry. Newborn screening. Diet/treatment.
Description	This video is a talk show-style interview with the parents of a child diagnosed with tyrosinemia type I and a genetic metabolic expert. The clip showcases the family's journey from newborn screening to the diagnosis to the creation of a disease foundation. A physician expert provides some scientific background on the disease and its treatment, and discusses the role of newborn screening in identifying patients.
Length (min:sec)	8:27
Speaker(s) Background	Parents, patient. Metabolic geneticist. Talk show host.
Objectives	1. Review the symptoms of tyrosinemia type 1. 2. Detail the diagnostic journey of a family impacted by a rare metabolic disease, and the role of newborn screening in getting to a diagnosis. 3. Describe the dietary treatment for this disease.
Educational utility	Provides a personal narrative about living with the disease but would be more effective if accompanied by some medical teaching.
Technical aspects	Excellent audio and 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	Promotes the Network of Tyrosinemia Advocates (NOTA). This video does not otherwise promote a commercial product, treatment, and/or medical device.
Source	The Balancing Act (Lifetime TV).

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