

## NPTR VIDEO CATALOG ENTRY

Disease	Tyrosinemia type I. Hepatorenal tyrosinemia. Amino acid disorders.
Title	Newborn Screening Not Always Enough for Detecting Tyrosinemia Type 1.
Link	<a href="https://youtu.be/PP-42VIQUFk">https://youtu.be/PP-42VIQUFk</a>
Key words	Tyrosinemia type I. Hepatorenal tyrosinemia. Amino acid disorders. Patient/family experience. Symptoms.
Description	In this video, the mother of Evan (born with tyrosinemia type I) details her family's experience in the newborn period. Evan was very ill and the journey to a diagnosis included advocacy and good luck. The particular issue with the negative newborn screen is unclear (e.g., false negative?, inadequate testing method?). It is a reminder that newborn screening is not infallible; even if a disease was tested for through newborn screening and found to be negative, medical providers should repeat testing if clinically indicated.
Length (min:sec)	3:11
Speaker(s) Background	Mother. Evan.
Objectives	1. Detail the symptoms of neonatal tyrosinemia type I.
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 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 does not promote a commercial product, treatment, and/or medical device.
Source	Rare Disease Report.
Author Date	MK. 11/12/2020

