

NPTR VIDEO CATALOG ENTRY

Disease	Homocystinuria (HCU). Amino acid disorders.
Title	Ethan's story - Living with HCU.
Link	https://youtu.be/g-QrsUni5Hk
Key words	Homocystinuria (HCU). Amino acid disorders. Patient/family experience. General disease overview. Symptoms. Pathophysiology. Biochemistry. Newborn screening. Diet/treatment. Prognosis.
Description	In this video, Ethan's parents describe their experience in finding out about his homocystinuria through newborn screening. A metabolic expert gives a general overview of the phenotype and the underlying pathobiochemistry. He discusses the dietary approach and the parents comment on the challenges of dealing with the disorder.
Length (min:sec)	8:21
Speaker Background	Parents. An expert in metabolic disease.
Objectives	<ol style="list-style-type: none"> 1. Discuss the phenotype of homocystinuria and the underlying pathophysiology/biochemistry. 2. Describe some of the challenges/experiences of patients with homocystinuria. 3. Discuss an overview of therapy for this disorder.
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 features	Good video and sound 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	The patient mentions certain treatment products. This video does not otherwise promote a commercial product, treatment, and/or medical device.

Source	NIHRtv - NIHR CLAHRC South Yorkshire.
Author	JA. 08/07/20