

### NPTR VIDEO CATALOG ENTRY

Disease	Homocystinuria (HCU). Amino acid disorders.
Title	HCU - Stacey's story.
Link	<a href="https://www.youtube.com/watch?v=ktm-yRF1I5E&amp;list=PLvz3MWdu8TP3wTE9wYNxq3WOxQxMbx4XV&amp;index=10">https://www.youtube.com/watch?v=ktm-yRF1I5E&amp;list=PLvz3MWdu8TP3wTE9wYNxq3WOxQxMbx4XV&amp;index=10</a>
Key words	Homocystinuria (HCU). Amino acid disorders. Patient/family experience. General disease overview. Symptoms. Pathophysiology. Biochemistry. Newborn screening. Diet/treatment.
Description	Throughout this video, Stacey discusses her experience growing up with homocystinuria and how it has affected her family and her lifestyle. Two metabolic experts provide a medical perspective of the disease and how it impacts management of the patient. The importance of newborn screening in identifying affected babies so treatment can begin is emphasized.
Length (min:sec)	5:35
Speaker Background	Patient. Parents. Two experts in metabolic disease.
Objectives	<ol style="list-style-type: none"> <li>1. Detail some of the symptoms/complications of homocystinuria and the underlying pathophysiology/biochemistry.</li> <li>2. Describe some of the challenges/experiences of patients with homocystinuria.</li> <li>3. Discuss an overview of therapy for this disorder.</li> </ol>
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	This video does not promote a commercial product, treatment, and/or medical device.

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