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Atypical infantile presentation of HCM detected incidentally by POCUS in Pediatric practitioner office-based setting

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Background: Point of care ultrasound (POCUS) modality offers a means of improving access to diagnosis and appropriate early treatment to achieve better quality of life.

POCUS is increasingly being performed by pediatric primary care in office-based practice and has applications throughout the spectrum of different pathologies, including cardiac applications (acute illness, pericardiac effusion, ventricular function and post-operative follow-up).

The skill is relatively easy acquired. However, the lack of exposure of this focused approach in most pediatric training programs in Italy remains a major obstacle.

Material and methods: Observational case of three months old infant presented with unexplained LV wall thickness detected incidentally by cardiac POCUS, which subsequently revealed to have Pompe disease.

POCUS cardiac examination performed by primary care pediatric practitioner in ambulatory-based setting practice.

Ventricular dimensions were obtained in the cardiac 4 chamber view (m-mode & 2 D recording).

Conclusions: Hypertrophic cardiomyopathy (HCM) is a genetically heterogeneous disorder with a large number of genes involved in disease causation (200 mutations in 10 genes).

Storage disorders and metabolic defects predominate in childhood HCM, and usually are recessive genetic defects, therefore, it is very important recognize particular echo-features of each HCM phenotype in order to plan the correct treatment and to improve patients' quality of life and survival.

POCUS in HCM setting may be used to enhance patient decision making regarding pursuit of targeted genetic panel testing.

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