# INTISARI

**Latar Belakang**: Propionic acidemia (PA) adalah gangguan metabolik langka yang diturunkan secara autosomal resesif akibat defisiensi enzim propionyl-CoA carboxylase, menyebabkan akumulasi metabolit toksik. Penatalaksanaan diet, termasuk pembatasan asupan protein dan penggunaan formula medis khusus, sangat penting. Namun, di daerah dengan sumber daya terbatas seperti Indonesia, kendala finansial dan keterbatasan akses terhadap formula tersebut menyulitkan penatalaksanaan.

**Tujuan**: Laporan kasus ini menyoroti tantangan dalam menangani seorang anak perempuan berusia 2 tahun dengan PA di Indonesia, dengan fokus pada dampak keterbatasan sumber daya terhadap manajemen nutrisi dan perawatan medisnya.

**Laporan Kasus:** Seorang anak perempuan berusia 1 tahun 5 bulan datang dengan gagal napas, kejang, dan asidosis metabolik berat. Pemeriksaan genetik mengonfirmasi PA. Penatalaksanaan awal meliputi penghentian asupan protein, hidrasi, dan pemberian levocarnitin. Perawatan jangka panjang melibatkan diet protein terkontrol, levocarnitin, dan natrium bikarbonat. Kendala finansial menyebabkan penghentian formula medis khusus, memicu kejang berulang dan peningkatan kadar amonia.

**Hasil dan Pembahasan**: Status gizi pasien membaik dari malnutrisi menjadi normal, dengan kemajuan signifikan dalam pertumbuhan dan perkembangan. Namun, perkembangan bahasa tetap stagnan, dan epilepsi memburuk, memerlukan dua obat anti-epilepsi. Keterbatasan akses terhadap formula medis khusus menghambat optimasi jangka panjang, meningkatkan risiko akumulasi metabolit toksik. Protein nabati, meskipun bermanfaat, berisiko menyebabkan defisiensi asam amino esensial, menekankan pentingnya pemantauan ketat dan edukasi keluarga.

**Kesimpulan**: Penatalaksanaan PA di daerah dengan sumber daya terbatas memerlukan pendekatan multidisiplin, termasuk manajemen diet, intervensi farmakologis, dan edukasi keluarga. Dukungan pemerintah, seperti subsidi dan peningkatan cakupan kesehatan, sangat penting untuk perawatan berkelanjutan dan hasil yang lebih baik.

**Kata Kunci**: Propionic acidemia, gangguan metabolik bawaan, gangguan metabolik

# ABSTRACT

**Background**: Propionic acidemia (PA) is a rare autosomal recessive metabolic disorder caused by propionyl-CoA carboxylase deficiency, leading to toxic metabolite accumulation. Dietary management, including restricted protein intake and specialized medical formulas, is critical. However, in resource-limited settings like Indonesia, financial constraints and limited access to these formulas complicate management.

**Objective**: This case report highlights the challenges in managing a 2-year-old girl with PA in Indonesia, focusing on the impact of limited resources on her nutritional and medical care.

**Case Report**: A 1-year-5-month-old girl presented with respiratory failure, seizures, and severe metabolic acidosis. Genetic testing confirmed PA. Initial management included protein cessation, hydration, and levocarnitine. Long-term care involved a controlled protein diet, levocarnitine, and sodium bicarbonate. Financial constraints led to discontinuation of medical food, causing recurrent seizures and elevated ammonia levels.

**Results and Discussion**: Nutritional status improved from malnutrition to normal, with significant growth and developmental progress. However, language development stagnated, and epilepsy worsened, requiring two anti-epileptic medications. Limited access to specialized medical foods hindered long-term optimization, increasing the risk of toxic metabolite accumulation. Plant-based protein, while beneficial, posed risks of amino acid deficiencies, emphasizing the need for strict monitoring and family education.

**Conclusion**: Managing PA in resource-limited settings requires a multidisciplinary approach, including dietary management, pharmacological interventions, and family education. Government support, such as subsidies and improved healthcare coverage, is essential for sustainable care and better outcomes.

**Keywords:** Propionic acidemia, inborn errors of metabolism, metabolic disorder