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MONOGENIC DIABETES IN 2 YEARS OLD GIRL: IS IT DEND?

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Background:
Monogenic forms of diabetes are still rare and not well understood. Their prevalence accounting for 1–4% of pediatric diabetes cases. Several genes encoding proteins important to β-cell function or regulation have been identified that lead to monogenic diabetes. However, awareness of these conditions may be lacking, and screening for them genetically is not routinely undertaken due to lack facility.

Objective:
To report a case diagnosis and management of monogenic diabetes and DEND in 2 years and 4 months old girl

Case:
Two years four months old girl, BW 8.4 kg (< -3 SD), BH 79 cm (< -3 SD), came to emergency department of Dr. Soetomo General Hospital. She referred from private hospital with DKA, pneumonia, seizure and loss of consciousness (GCS 113). In the past 2 months she has polydipsia, polyuria, polyphagia and loss of weight. Abdominal pain and difficulty of breathing were found in the past 4 days before admission. She has developmental delay such as motoric delay, speech delay and has involuntary movement too. Laboratory examination revealed BS 477 mg/dL; pH 7.32; HCO3 9.8; ketone 4.8; HbA1c 4.7; c-peptide 1.35 (1.1-4.4 ng/ml). Treatment with Glibenclamid and novorapid can control BS within normal limit.

Conclusion:
The girl was diagnosed DEND using criteria developmental delay, epilepsy and diabetes. To confirm diagnosis of DEND needs genetic mutation testing.
Keywords:
Monogenic diabetes, DKA, DEND

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