The challenges faced in the management of Congenital Generalized Lipodystrophy

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Background

- Congenital Generalized Lipodystrophy (CGL) is a rare autosomal recessive disease with around 500 cases worldwide.1
- Absence of functional adipocytes results in storage of lipids in other tissues, e.g. muscle & liver.1,2
- Metrelin injections are beneficial in lowering triglycerides (TG), liver enzymes, and better glycemic control.2,3 Unfortunately, it is not available in Egypt.
- We hereby report 2 cases diagnosed with CGL, and the challenges faced in their management in the absence of metrelin.

Case summary

Case 1

- A 2-year-old girl born to non-consanguineous parents presented with facial dysmorphism and loss of subcutaneous (SC) fat.
- At the age of 8 years, she had recurrent attacks of abdominal pain. Ultrasonography showed bilateral ovarian cyst torsion, and she had oophorectomy, and appendectomy.
- She was diagnosed clinically in our hospital with CGL at the age of 9 years. Her TG, fasting blood glucose (FBG), HOMA-IR and liver functions were normal.
- After skipping follow up for 2 years, xanthomas were observed on the limbs with severe hyperTG (13,000 mg/dl). She was also diagnosed with DM (HbA1c 13%) and discharged on dietary advice, insulin, metformin, omega-3, fenofibrates & statins.
- At the age of 12 years, she complained of severe abdominal pain with shooting serum lipase, and admitted with acute pancreatitis due to hyperTG. CT abdomen with contrast showed pseudopancreatic cyst; which was operated.
- She continued to have recurrent attacks of pancreatitis, despite that antilipidemic treatment was maximized. Metformin had to be stopped as it was suspected to be a risk factor for pancreatitis. Her last HbA1c is 9.6% on insulin requirement 5 U/Kg/day.

Case 2

- A 12-year-old girl born to consanguineous parents was diagnosed with CGL at the age of 4 months with typical facial features, hepatomegaly, hypertrichosis, hyperTG, and elevated liver enzymes, with normal OGTT. Her liver biopsy showed moderate macronuclear steatosis and cirrhosis. She was given fenofibrates and statins.
- At the age of 4.5 years, she presented with precocious puberty [pubic hair (Tanner 2), breast (Tanner 1)]. Her height was 119 cm (+2.6SD), growth velocity +5SD, with advanced bone age. Her labs showed mildly elevated DHEA-S, normal testosterone, and 17-OH progesterone levels and was diagnosed with premature adrenarche.
- She was diagnosed with DM at age 7 years (HOMA-IR 8.1%, HbA1c 8.4%). She has good glycemic control on dietary modification, & metformin.
- Her echocardiography showed dilated left ventricle, depressed systolic function.

Conclusion

- Patients with CGL can be diagnosed clinically as their phenotypic characteristics are unique; however, different presentations and severity exist.
- Without metrelin, challenges are faced affecting the quality of life and treatment should be individualized.

References


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