lissencephaly, arthrogryposis, seizures, ventilator dependent due to respiratory failure and then discharged on home ventilator and anti-epileptics. Other medical disorders: included dysphagia, gastro-esophageal reflux disease, tube feeding, tracheostomy and ventilator dependence, intermittent sinus bradycardia. He was diagnosed at the age of 4 years with type 1 diabetes mellitus presented with hyperglycemic hyperosmolar state. (Genetic test: **SMPD4 variant** Homozygous2: g.130931103C>A;c.370G>T;p.E124).

**Conclusion** SMPD4 regulates mitotic nuclear envelope dynamics and its loss causes neurodevelopmental abnormalities and diabetes

## 38 MANAGING SEVERE DIABETES IN CONGENITAL LIPODYSTROPHY: A CASE REPORT

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**Background** Congenital lipodystrophy is an exceedingly rare genetic disorder with an estimated worldwide prevalence of less than 1 case per million individuals. Four distinct genetic mutations underlie congenital lipodystrophy, including AGPAT2, BSCL2, CAV1, and PTRF genes. This condition is Characterized by the near absence of subcutaneous adipose tissue, resulting in fat deposition in internal organs such as the liver. It leads to cirrhosis and metabolic disturbances, including hypertriglyceridemia and insulin resistance, often precipitating early-onset diabetes mellitus. We report a case of congenital lipodystrophy with a focus on the management of severe diabetes with insulin resistance.

**Case Report(s)** Our patient is a 14-year-old female with congenital lipodystrophy, initially manifesting as hypertriglyceridemia with normal blood glucose levels. Subsequently, at 14 years of age she experienced irregular menstrual periods and worsening acanthosis nigricans. Continuous glucose monitoring over two weeks revealed consistently elevated fasting and postprandial blood glucose readings. Physical examination demonstrated a muscular appearance, generalized lipoatrophy, enlarged hands and feet, hirsutism, and prominent acanthosis nigricans on the neck and flexor folds. Laboratory findings indicated an elevated hemoglobin A1c of 7.7% and hypertriglyceridemia.

Treatment was initiated with insulin, metformin, and dietary modifications, eventually necessitating high insulin doses, including insulin Lantus (60 Units daily) and insulin Lispro (35 Units with meals thrice daily). Subsequently, metreleptin therapy was introduced, resulting in improved glycemic control, with blood glucose levels stabilizing between 70 and 150 mg/dl, allowing for the discontinuation of insulin therapy.

**Conclusion(s)** Congenital lipodystrophy poses a rare and complex challenge, often complicated by severe diabetes due to insulin resistance. Metreleptin emerges as an effective therapeutic option for managing diabetes in these patients. As a recombinant analog of the human hormone leptin, metreleptin offers promising outcomes in glycemic control and warrants consideration in the management of congenital lipodystrophy-related diabetes.

## 39 A DONHUE'S SYNDROME CASE : RARE SEVERE SYNDROMIC INSULINE RESISTANCE

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**Background** Donohue Syndrome is the most severe form of the rare genetic severe insulin resistance syndromes caused by mutation in the insulin gene receptor (INSR) with a lifespan under 2 years mainly because of intercurrent infections.

Resulting from a homozygous or compound heterozygous mutations of the INSR (INSR; 19p13.3– p13.2). It associates severe growth retardation beginning prenatally, hyperinsulinism with hyperglycemia and dysmorphic features.We report this case due to the extreme rarity of the syndrome and to present its clinical, biological findings and the underlying genetics.

**Case Report(s)** The case is an Algerian female infant born to consanguineous parents via caesarian section at 37weeks gestation due to severe intrauterine growth restriction.

She was admitted in our department at 4months old showing septic signs (high fever, tachycardia) with severe growth stunt (over -7DS), lipoatrophy, cholestasis, abdominal distension and dysmorphic features: craniofacial abnormalities with elfin facies and large low set ears, hypertrichosis and clitoromegaly.

Laboratory results showed hyperinsulinism with high insulin, peptic C and fasting hypoglycemia; postprandial hyperglycemia ; cholestatic hyperbilirubinemia and low triglyceride and cholesterol levels. C-reactive protein was high.

This presentation suggested DS which was confirmed by finding of a homozygous deletion in 19 p13.2 encompassing exon 2 of INSR.

Antibiotics were immediately put and growth hormone therapy was initiated (0,35 mg/kg/d); unfortunately, 3 days later she died from apparent septicemia. Our patient had no opportunity for treatment with recombinant IGF-1.

**Conclusion(s)** Our case shows a classic presentation of DS, with a fatal prognosis. Treatment is only symptomatic and requires a multidisciplinary team. Recombinant IGF-1 shows promise but the benefit is yet to be established.

Majority of cases are found where consanguineous marriages prevail, thus genetic counseling and discouraging inbreeding should be done as preventive measures awaiting optimal treatment protocol.

## 40 TYPE 1 DIABETES MELLITUS AND AUTOIMMUNITY

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**Background** We conducted a retrospective study in the Diabetology and Nutrition Endocrinology Department of the Mohammed VI University Hospital in Oujda, Morocco. For this, we used SPSS 21 software for data collection and analysis.