Background: Cox-deficient Leigh syndrome French Canadian variant characterized by hypotonia, developmental delay, mild facial dysmorphism, chronic well-compensated metabolic acidosis, episodes of severe acidosis, and coma associated with high mortality. Here we presented a patient with LRPPRC to discuss her clinical and laboratory findings.

Case Report:
- 17 years old female patient.
- 38 weeks of gestation, NSVY delivery
- Consanguinity in parents was from the same village
- Delay in reaching language and motor skills milestones
- At 4 years old, loss of acquired skill
- At 11 years old, neurodevelopmental delay, muscular hypotonia, truncal ataxia
- At 13 years old, general weakness, gaze palsy, and distal spasticity

Physical Examination:
- Weight: 43 kg (−2.7 SDS)
- Height: 157 cm (-1.02 SDS)
- BMI: 17.4 (-2.7 SDS)
- Head Circumstance: 56 cm (-0.05 SDS)
- Ataxia+, Dysarthria+, Dysmetria+/+
- Biletaeral hand and finger spasticity
- Walk with an ankle-foot orthosis.

Laboratory Findings:
- CSF lactate elevation (25.1 mg/dL) and blood lactate was elevated (27 mg/dl).
- The cranial MRI: symmetrical signal changes in brain stem and cerebellum and MRS was normal.
- EMG: sensorimotor axonal demyelinating polyneuropathy
- LRPPRC gene: Homozygous mutation c.2726_2728del (p.K909del)

Results:
Even in the absence of any metabolic crisis, LRPPRC mutation should be considered in children with infantile or early childhood neurodegenerative disorders. Although severe or even fatal lactic acidosis has been reported in the literature, we did not detect decompensation in our patient, except for a mild lactate elevation. On the contrary to the literature, the results of our patient appeared as a mild variant. The ketogenic diet may be the option of treatment.