BERBERINE TREATMENT IN COCKAYNE SYNDROME

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Cockayne syndrome (CS) is a rare autosomal recessive genetic disorder associated with premature aging. It is one of the progeroid syndrome in congenital lipodystrophies.

- Its classical features are defined as microcephaly, cataract, lipodystrophy, hearing loss, progressive loss of adipose tissue, contractures in extremities.
- In addition to these features, cases with very high insulin levels and insulin resistance have also been reported. Berberine, the main active ingredient of an ancient Chinese herb Coptis chinensis French, has been used in the treatment of type 2 diabetes for insulin resistance.

A 14-year-old male patient, presented with joint contractures apparent in the lower extremities and subcutaneous adipose tissue loss. He had an atypical facial appearance with a thin nose, large ears, sunken eyes. He also had cataract, hearing loss and microcephaly.

On the cranial computer tomography scan ventriculomegaly and calcifications in the basal ganglia.

Acro-osteolysis in the distal phalanges.

His whole exom sequencing was held and a homozygous mutation in COCANE-A gene was detected. Fasting blood glucose measurements were between 200-300 mg/dl. Fasting insulin level was determined as 250 mg/dl. A daily dose of 500 mg berberine was started. In the follow-up of the patient, blood sugars came to normal ranges, fasting insulin level came to 90-100 mg/dl range. Cockayne syndrome is a rare incurable genetic disorder with insulin resistance. Berberine may be an option in the treatment of insulin resistance of the syndrome. More scientific research and studies are needed.