CLINICAL IMAGE IN MEDICAL PRACTICE

LAURENCE - MOON - BIEDL SYNDROME

ISLAM QT¹, ISLAM QT²

A 23-year-old lady, known diabetic and hypertensive, presented with complaints of visual impairment and obesity. On further enquiry she was found to have obesity problem and learning difficulties since childhood. On examination patient was found to have nystagmus, polydactyly and fundoscopic examination revealed retinitis pigmentosa and cataract left eye. The patient was clinically suspected having a rare autosomal recessive disorder, the Laurence-Moon-Biedl syndrome (LMBS).

The LMBS affects approximately 1/100,000 births. Genes on chromosomes 16, 11, 3, 15, and 20 have been associated with LMBS.³ The possible hypothesis of pathogenesis is leptin resistance which causes hypothalamic dysfunction resulting in loss of satiety and decreased GnRH release finally leading to obesity and primary hypogonadism.

Secondary symptoms may also occur, such as delayed development, speech problems,⁴ Type 2 diabetes and ataxia were present in this patient.

For diagnosis, four primary symptoms should be present, or three primary plus two secondary symptoms. There is no specific test to detect the presence of LMBS,²

Fig: Young, very obese patient, Laurence - Moon - Biedl syndrome

Reference: