PAEDIATRIC DERMATOLOGY

DERMAL ERYTHROPOIESIS WITH NON KETOTIC HYPERGLYCINEMIA : A RARE CASE REPORT OF BLUEBERRY MUFFIN BABY

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BACKGROUND : Blueberry Muffin Baby is term given to characteristic eruption in neonates, often present at birth, comprising widespread, purple, erythematous, oval or circular macules, papules and nodules reflecting dermal erythropoiesis seen in a number of congenital infections, notably rubella, cytomegalovirus, coxsackie B2 infection, parvovirus B19, congenital syphilis, toxoplasmosis, rhesus incompatibility.
Nonketotic Hyperglycinemia (NKHG) is an inborn error of glycine degradation in which large quantities of glycine accumulate in body tissues, including CNS, and has been associated with “Blue-Berry Muffin” Baby.

OBSERVATION : A 1½ month-old-male child, born out of a consanguineous marriage, to Gravida 5,Para 3,Abortion 1 with live 1,female with history of Rh incompability, presented with complaints of asymptomatic dark lesions over the right arm, back, left thigh and face since birth, with history of myoclonic jerks and tachypnoea.
On examination: Multiple asymptomatic Erythematous to Dusky blue, non blanchable domed papules present over the arm and over face and back.
Complete blood counts: Hb 10.4 g%; TLC: 6500; DLC: N5 L81 M8 E6 B0 Platelet Counts 512,000; PS: Normochromic Blood Picture with neutropenia with thrombocytosis. Serum.Ammonia :81 mcg/dl
Carnitine/Acyl Carnitine profile showed Normal total carnitine, low free carnitine, very low free/acyl carnitine ratio.

KEY MESSAGE : The blueberry muffin baby has been associated historically with congenital viral infections and hematologic dyscrasias. We report this case because of its rare association with Nonketotic Hyperglycinemia(NKHG)