

Early diagnosis of phenylketonuria. Follow up of 2 cases Diagnostico precoz de fenilquetonuria. Seguimiento de 2 casos.

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A screening program for the early diagnosis of phenylketonuria was initiated 24 months ago in the Servicio de Salud Metropolitano Central. Since then, 2 cases with phenylketonuria have been early diagnosed. These patients started their nutritional treatment, consisting of a phenylalanine restricted diet at 14 and 17 days of age. The children are submitted to periodic medical, anthropometric, neurologic, biochemical and psychometric analysis. With a strict control they maintain phenylalanine plasma levels between 2 and 6 mg%. Both patients have a normal psychomotor development at 4.5 and 6.5 months of age and an anthropometric development at p50 of NCHS.