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NEONATAL SCREENING – SHORT REPORT

Long-term follow-up study of patients with phenylketonuria detected by the newborn screening programme in Japan

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Summary The objective of this report is to determine the need for and value of the long-term follow-up study of phenylketonuria (PKU) patients detected by newborn screening (NBS) in Japan. NBS was started in 1977 and the nationwide follow-up study of the identified patients was introduced into the NBS system. Outcome data from the continuous follow-up study showed in 1993 that IQ of PKU patients was inversely correlated with blood phenylalanine levels. Accordingly, in 1995, new treatment guidelines were issued that involved more stringent restriction of phenylalanine levels. Follow-up data confirmed that mean blood phenylalanine levels decreased after the introduction of the new guidelines, which included the recommendation to start dietary treatment within 20 days *postpartum*. Follow-up data also confirmed that dietary treatment did in fact commence earlier after the guidelines were issued. The need for lifelong dietary treatment is a difficult issue and the number of patients who stop dietary treatment was found to increase gradually with age. At present 60% of PKU patients born between 1977 and 1981 have stopped their dietary restriction of phenylalanine. The data gained from NBS and the long-term follow-up study were found to be valuable for the improvement of blood phenylalanine levels for patients with PKU, indicating the need for parties responsible for NBS and the follow-up study of the identified patients to work cooperatively. Further, the evaluations of the effectiveness of the two initiatives as well as the treatment guidelines issued should be based on outcome data, which depend on the continuation of the follow-up study of patients with PKU.

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