The impact of Phenylketonuria (PKU) is particularly critical during pregnancy, as elevated and prolonged intrauterine Phe levels have teratogenic effects to the developing fetus; this condition, known as ‘maternal PKU syndrome’, can cause various deficits including developmental delay, low birth weight, microcephaly, congenital heart disease, craniofacial dysmorphism, and intellectual impairment. 3

For this reason, women with PKU who are planning to have a child should follow a strict diet before conception and continue it throughout gestation, to keep Phe concentrations at safe levels. Granules coated with functional additives, the content of the granules is gradually released in the small intestine, making the product tasteless and odorless, and allowing it to overcome practical problems associated with free amino acids, such as bitter taste, unpleasant smell, aftertaste and constipation.

Thanks to the special coating granules remain intact during consumption, masking amino acid odour and taste and preventing aftertaste. This allows an easier palatability.

We present the case of a young Italian woman (1985) with PKU who grew up without following any diet during childhood. She showed no particular problems apart from frequent abdominal pain, irritability, and delayed height development. In adulthood, she led a normal life until she became pregnant: after some miscarriages, she was diagnosed with classic PKU at the age of 30 (blood Phe concentration = 1812.46 µmol/L [29.94 mg/dl]). Subsequently, she started a strict dietary regimen with controlled Phe intake (97.28 mg daily) supplemented with two protein substitutes: 33 g daily of a slow-release large neutral amino acids (LNAA) preparation; a ready-to-drink product containing essential and non-essential amino acids, carbohydrates, vitamins, and minerals. This diet allowed her to achieve optimal metabolic control, reducing her blood concentration of Phe to levels that did not interfere with a possible pregnancy. When she became pregnant at the age of 30 (blood Phe concentration = 1812.46 µmol/L [29.94 mg/dl]), she started taking (31.5g daily) a prolonged-release amino acid mixture with vitamins, minerals and other nutrients (PKU Golike 16+ APR Applied Pharma Research s.a). This product has been selected because many clinical studies evidenced safety of the excipients for use also during pregnancy and lactation. 4-6

Unfortunately, the beginning of pregnancy coincided with the onset of a disabling gastroesophageal reflux, which prevented her from resting during the night. This led to reduced dietary compliance and, consequently, worsening metabolic control. Thereafter, she was prescribed a new protein replacement, and from 26 weeks gestation she started taking (31.5g daily) a prolonged-release amino acid mixture with vitamins, minerals and other nutrients (PKU Golike 16+ APR Applied Pharma Research s.a). This product has been selected because many clinical studies evidenced safety of the excipients for use also during pregnancy and lactation.4-6

The patient successfully carried to term her pregnancy with the spontaneous delivery of a healthy baby girl at 39 weeks. All auxological parameters of the newborn were within normal limits: birth weight = 2.73 kg; length = 48 cm; head circumference = 32 cm. After delivery, the mother continued to take the prolonged-release amino acid mixture with good adherence to dietary therapy and good metabolic control, which allowed her to breastfeed the child.

The patient continued to breastfeed the child.