In the novel *The Pickwick Papers*, author Charles Dickens describes a fat boy with red cheeks and a sleepy expression. The boy is characterized as a small ball, an insatiable eater, and a "boa constrictor." Such a description aptly captures the features of Prader-Willi syndrome (OMIM 176270) (Fig. 1).

In 1956, Swiss endocrinologists Prader, Labhart, and Willi described a group of nine unrelated children who shared certain physical traits: short stature, obesity, hypotonia, incomplete sexual development, and mild intellectual disability (Prader et al., 1956a; Prader et al., 1956b). This cluster of symptoms was later named Prader-Willi syndrome (PWS). The clinical symptomatology of PWS changes markedly as the child develops. Theoretically, we can distinguish the fetal and neonatal stage, early childhood, late childhood, adolescence, and adulthood.

Pregnancy usually proceeds normally, although reduced fetal movement may be felt by the mother prenatally. The newborn is typically noticeably hypotonic, has a short birth length, low weight, and a weak cry. Weight gain is inadequate due to feeding difficulties caused by a weak sucking reflex. Hypogenitalism and thermoregulation disorders are also often present. The physical signs of PWS are not always immediately striking in all children, and some symptoms may appear earlier or later. In differential diagnosis during this period, assessing facial dysmorphism can be helpful, as certain characteristic features may already be present (narrow forehead, dolichocephaly, almond-shaped eye slits, thin upper lip with downturned corners—“fish mouth,” small receding lower jaw, narrow nose, low-set small ears, hypopigmentation). However, these symptoms become significantly more pronounced with age. Nearly half of the children exhibit strabismus. Skin syndactyly of the fingers and clinodactyly of the fifth finger are also frequently observed. During the first year, muscle hypotonia partly improves, motor activity increases, and the phenotype becomes more evident.