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VERSO IL CLIL
Genetics: from peas to diseases

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The great contribution of Gregor Mendel, the «father of genetics», was to show that inheritance follows predictable patterns. His experimental subject was very ordinary (the common garden pea, *Pisum sativum*) and the traits he selected for study were unremarkable: flower colour and seed shape, for example. But the principles he discovered opened the flood-gates for the studies that have come to be known as classical genetics.

Mendel's discoveries have important modern applications in medical genetics, the study of inherited disorders. One human disorder that follows a Mendelian pattern of inheritance is Huntington's disease, which causes a progressive destruction of brain cells, ending in death within 10 to 20 years of the onset of symptoms. Most human hereditary disorders are evident at birth or in early infancy, but Huntington's usually first manifests itself among individuals in their 30s and 40s. One consequence of this late onset is that the individual often already has children who, in turn, carry the same hidden trait. According to Mendelian principles, any child of a man or woman who develops Huntington's has a fifty-fifty chance of also developing the disorder.

Researches into Huntington's disease have led geneticists to the shores of Lake Maracaibo in Venezuela. Among the inhabitants of a remote village, reachable only by small fishing boats, are more than 100 individuals with Huntington's and another 1000 who are at high risk for the disorder. All are descendants of a single woman, Maria Concepción Soto: she lived in the early nineteenth century and is thought to have inherited the disorder from a German sailor who was her father.

In recent years, geneticists have compiled a pedigree (a detailed chart) tracing the family relationships and incidence of Huntington's among almost 10 000 individuals. This pedigree has made it possible to predict which children in the population are at a high risk of eventually developing the disease.

A close up shot of sweet pea flowers.

A Why are Mendel and his discoveries so important for genetics?
B At what age can Huntington's disease manifest itself?