

Research

Human Genetics & Embryology

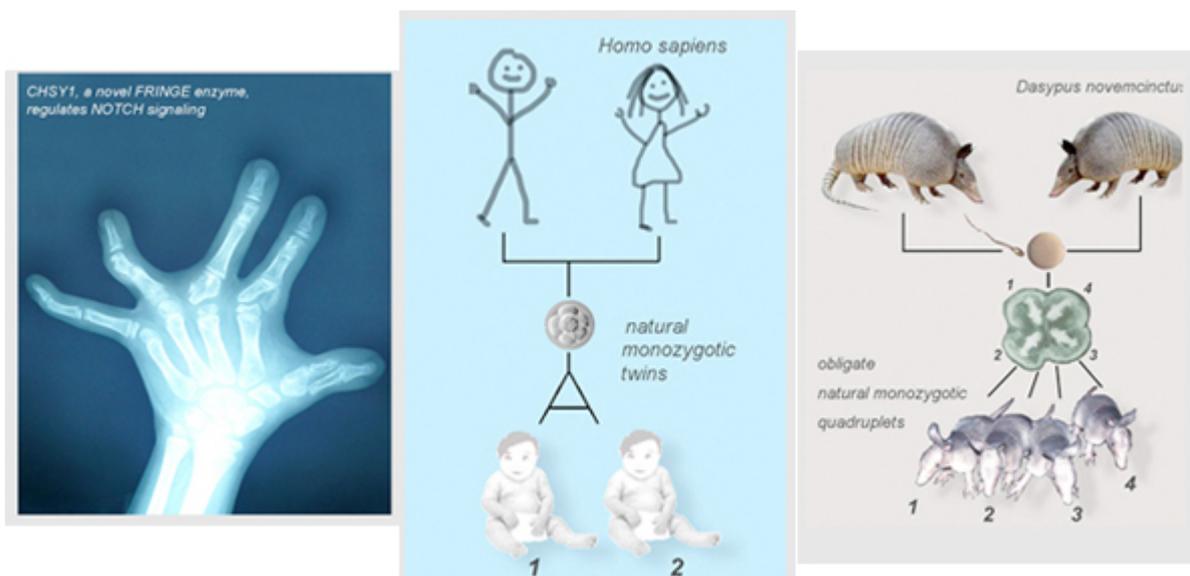
The laboratory of Dr. Bruno Reversade investigates human embryonic development and its associated genetic diseases. By addressing rare human pedigrees his group examines inherited disorders that produce birth defects or remarkable events such as the making of identical twins.

To understand the principles that govern normal embryogenesis in the human species, our lab researches genetic diseases that cause congenital birth defects or unusual phenotypes.

We take advantage of rare human pedigrees from highly consanguineous populations to characterize novel genes whose mutations affect the development of the human embryo. Following genetic mapping and high throughput sequencing, we undertake a functional analysis by using animal models (such as flies, frogs, fish and mice) and tools from molecular biology and biochemistry. In so doing, we hope to gain a clear mechanistic explanation of the fundamental processes underlying disease states and normal human embryonic development.

For example we have identified PYCR1 as causing an autosomal recessive form of premature ageing and CHSY1 as a novel NOTCH modulator essential for limb patterning.

For more information, please visit the lab's website at www.reversade.com or contact [Bruno Reversade](#).



We have a particular interest in unraveling the genetic basis of identical twinning in humans. Identical twins also referred to as monozygotic (MZ) arise once every 300 births when a single human embryo splits in two giving birth to a pair of monoclonal babies. Ongoing work, based on families with multiple incidences of MZ twins suggests that the birth of natural human reproductive clones may be under genetic control.