

Zellweger spectrum disorders are a group of rare, genetic, multisystem disorders that were once thought to be separate entities. These disorders are now classified as different expressions (variants) of one disease process. Collectively, they form a spectrum or continuum of disease. Zellweger syndrome is the most severe form; neonatal adrenoleukodystrophy is the intermediate form; and infantile Refsum disease is the mildest form. Zellweger spectrum disorders can affect most organs of the body. Neurological deficits, loss of muscle tone (hypotonia), hearing loss, vision problems, liver dysfunction, and kidney abnormalities are common findings. Zellweger spectrum disorders often result in severe, life-threatening complications early during infancy. Some individuals with milder forms have lived into adulthood. Zellweger spectrum disorders are inherited as autosomal recessive traits. Zellweger spectrum disorders are also known as peroxisome biogenesis disorders (PBDs) - a group of disorders characterized by the failure of the body to produce peroxisomes that function properly. Peroxisomes are very small, membrane-bound structures within the gel-like fluid (cytoplasm) of cells that play a vital role in numerous biochemical processes in the body. PBDs are subdivided into the three Zellweger spectrum disorders and rhizomelic chondrodysplasia punctata. Zellweger spectrum disorders are usually apparent at birth. They affect individuals of all ethnic groups. In the United States, the combined incidence of these disorders is at least 1 in 50,000 live births. Because some cases go undiagnosed, determining these disorders true frequency in the general population is difficult.