Zellweger Syndrome

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Zellweger Syndrome Information Page.

Zellweger syndrome is one of a group of four related diseases called peroxisome biogenesis disorders (PBD). The diseases are caused by defects in any one of 13 genes, termed PEX genes, required for the normal formation and function of peroxisomes. The PBDs are divided into two groups: Zellweger spectrum disorders and Rhizomelic Chondrodysplasia Punctua spectrum. The Zellweger spectrum is comprised of three disorders that have considerable overlap of features. These include Zellweger syndrome (ZS, the most severe form), neonatal adrenoleukodystrophy (NALD), and Infantile Refsum disease (IRD, the least severe form). Peroxisomes are cell structures that break down toxic substances and synthesize lipids (fatty acids, oils, and waxes) that are necessary for cell function. Peroxisomes are required for normal brain development and function and the formation of myelin, the whitish substance that coats nerve fibers. They are also required for normal eye, liver, kidney, and bone functions. Zellweger spectrum disorders result from dysfunctional lipid metabolism, including the over-accumulation of very long-chain fatty acids and phytanic acid, and defects of bile acids and plasmalogens--specialized lipids found in cell membranes and myelin sheaths of nerve fibers. Symptoms of these disorders include an enlarged liver; characteristic facial features such as a high forehead, underdeveloped eyebrow ridges, and wide-set eyes; and neurological abnormalities such as cognitive impairment and seizures. Infants will Zellweger syndrome also lack muscle tone, sometimes to the point of being unable to move, and may not be able to suck or swallow. Some babies will be born with glaucoma, retinal degeneration, and impaired hearing. Jaundice and gastrointestinal bleeding also may occur.