# AICARDI SYNDROME

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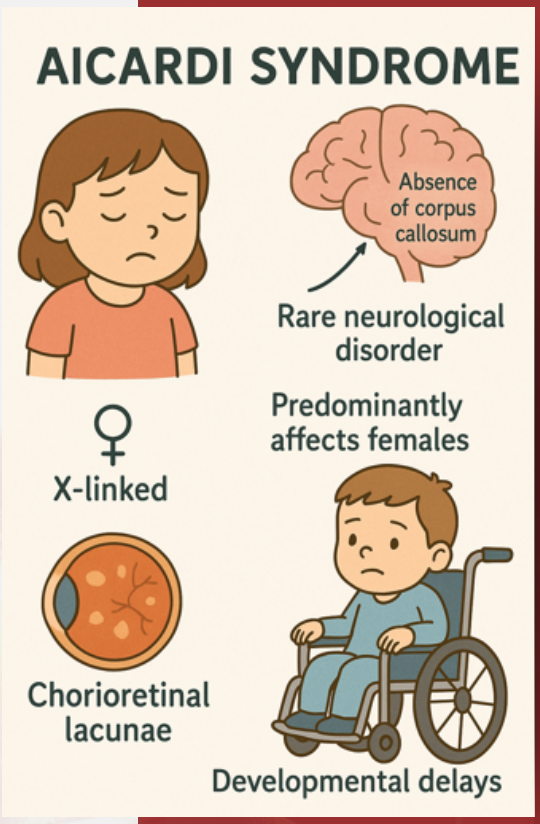
*Aicardi syndrome* is caused by abnormalities in genes linked to brain development. It is an X-linked dominant condition, predominantly affecting females. Males with the mutation are unlikely to survive. This rare congenital neurological condition occurs in one in 100,000 live births in the United States, one in 93,000 in the Netherlands, and one in 110,000 in Northern Ireland. Approximately 1,000 cases have been documented in the United States, with an estimated 4,000 cases reported globally. Currently, there is no comprehensive data or published research on the incidence or prevalence of Aicardi syndrome in Malaysia.




Aicardi syndrome is usually diagnosed in infancy and is characterised by several symptoms, including partial or complete absence of the corpus callosum—a structure that links the two brain hemispheres—leading to significant neurological difficulties. It is also marked by severe seizures in infants during the initial stages of life. Imaging tools such as EEG and MRI can be utilised to assess brain structure and seizure activity. Chorioretinal lacunae, which are distinctive retinal lesions, are a hallmark of the illness. Children affected by this syndrome often have developmental delays, including difficulties in social interactions, speech, and movement, since their cognitive and physical growth is commonly hindered. Spinal deformities, including scoliosis, are common in patients with Aicardi syndrome.

Individuals with Aicardi syndrome often experience developmental delays, including speech problems, which can lead to difficulties with verbal communication; some may be nonverbal. The life expectancy of people with Aicardi syndrome varies significantly depending on the severity of their symptoms and associated medical conditions. Factors such as the frequency and severity of seizures, the degree of brain abnormalities, and the presence of other issues like respiratory or heart conditions can impact their lifespan. Many children with Aicardi syndrome encounter significant health challenges during early life, including respiratory problems and seizures, which may result in a substantial proportion not surviving past early infancy.

However, some individuals with Aicardi syndrome have managed to survive into adolescence and beyond due to breakthroughs in medical care, notably in seizure management. It is projected that over 60% of those affected will live past the age of 10. While less common, there are cases of people with Aicardi syndrome living into their 20s and 30s. The probability of reaching the age of 27 is roughly 62%. An individual's lifespan is significantly influenced by supportive care, effective seizure management, and the treatment of additional issues.

Aicardi syndrome is incurable; therefore, treatment is primarily symptomatic, focusing on managing seizures, which are among the most significant and challenging aspects of the condition. Therapies are designed to alleviate symptoms and improve quality of life. These treatments include anti-epileptic drugs for seizure management and early intervention programmes to facilitate physical, occupational, and speech therapy in attaining developmental goals. Surgical intervention may be required in some cases to manage severe epilepsy or to rectify skeletal deformities. The retinal anomalies associated with the condition necessitate consistent surveillance and assistance for visual impairments.

Despite the numerous challenges, many individuals with Aicardi syndrome can lead fulfilling lives with early diagnosis, specialised treatment, and supportive therapy. Raising awareness of this rare condition equips families and healthcare providers with essential knowledge and resources for optimal management. To improve the quality of life for those affected by this rare genetic illness, it is essential to stay informed and advocate for ongoing research.