

Spinal Muscular Atrophy: Non-Curative Disease

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A one-month-old baby girl with spinal muscular atrophy type 1 with generalized muscle weakness is presented. She was fully investigated and diagnosed with spinal muscular atrophy type 1. She was managed conservatively and with supportive treatment. She died at the age of 13 months.

Spinal Muscular Atrophy (SMA) is a non-curative disease with a wide degree of clinical and genetic heterogeneity that we must take into consideration because of the serious prognosis. It targets the muscles which result in weakness and atrophy.

Several therapeutic approaches have been proposed and investigated. They are focused on increasing the availability of Survival of Motor Neuron (SMN) protein which has been found effective when tried in animal modelling studies.

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