

Table 1. Key investigation results in AA.

Investigation	Findings
Computed Tomography (CT) Brain	No acute intracranial haemorrhage. Basal ganglia calcification and generalised parenchymal atrophy.
Magnetic Resonance Imaging (MRI) Brain	Bilateral calcifications of the putamen on CT correlates with diffuse high T2 signal on MRI. No focal lesion is identified throughout. No diffusion restriction/infarct or venous thrombosis. Appearances of the generalised mild atrophy and basal ganglia calcifications remain non-specific, the underlying cause/condition uncertain.
Electroencephalogram (EEG)	Abnormal EEG. The background is dominated by diffuse high amplitude delta slowing with some intermixed theta. The background is poorly organised but not consistent with hypersarrhythmia but rather a moderate diffuse encephalopathy. There are no epileptiform discharges. Throughout the recording there are multiple clinical annotations of “tensing” as well as tongue sticking out. These do not have abnormal EEG correlate. However, there is a clinical episode of the baby outstretching arms that is associated with a decremental response on EEG and consistent with seizure that is likely infantile spasm.

Table 2. Key findings in Aicardi-Goutières Syndrome and TORCH Syndrome.

	Pseudo-TORCH Syndrome	TORCH Syndrome			
	Aicardi-Goutières Syndrome	Toxoplasmosis	Rubella	Cytomegalovirus (CMV)	Herpes Simplex Virus (HSV)
Aetiology	Autosomal dominant Autosomal recessive	Toxoplasmosis gondii	Rubella virus	Cytomegalovirus	Herpes Simplex Virus 2
Cardiac System	Cardiomyopathy	-	Patent ductus arteriosus Pulmonary artery stenosis Coarctation of aorta Myocarditis	-	Myocarditis
Respiratory System	-	Pneumonitis	Pneumonitis	Pneumonitis	-
Gastrointestinal System	Hepatosplenomegaly Jaundice	Hepatosplenomegaly Jaundice	Hepatosplenomegaly Jaundice	Hepatosplenomegaly Jaundice	Hepatosplenomegaly Jaundice Oral ulcerations
Endocrine System	Hypothyroidism Insulin dependent diabetes mellitus Micropenis	-	Hypothyroidism Insulin dependent diabetes mellitus	-	-

Nervous System	Basal Ganglia calcifications Cerebral atrophy Microcephaly Seizures Mental impairment Tetraplegia Dystonia Hypo/hypertonia Extrapyramidal signs Pyramidal signs Developmental regression	Hydrocephalus Microcephaly Intracranial calcifications Seizures	Hydrocephalus Microcephaly Mental impairment	Microcephaly Periventricular calcifications Hypotonia Seizures	Hydrocephalus Microcephaly Seizures Hypertonia
Musculoskeletal System	-	-	Cleft lip/palate Micrognathia	-	-
Skin	Petechiae Purpura Chilblain lesions Acrocyanosis Periungual erythema	Petechiae Purpura Maculopapular rash	Petechiae Purpura	Petechiae Purpura	Petechiae Purpura Vesicles
Vision	Impaired vision Nystagmus	Impaired vision Chorioretinitis	Impaired vision Chorioretinitis Cataracts Microphthalmia	Impaired vision Chorioretinitis	Impaired vision Chorioretinitis Cataracts
Hearing	-	-	Hearing deficits	Hearing deficits	Hearing deficits

Investigation Findings Suggestive of Diagnosis	Negative TORCH infection screen Elevated INF-alpha in the cerebrospinal fluid (CSF) Lymphocytosis in CSF Elevated neopterin and biopterins in CSF Positive genetic screen	Toxoplasmosis serology positive	Rubella serology positive	CMV serology positive	HSV 1 or 2 polymerase chain reaction (PCR) positive HSV1 or 2 serology positive
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