

The aetiologies of epilepsy

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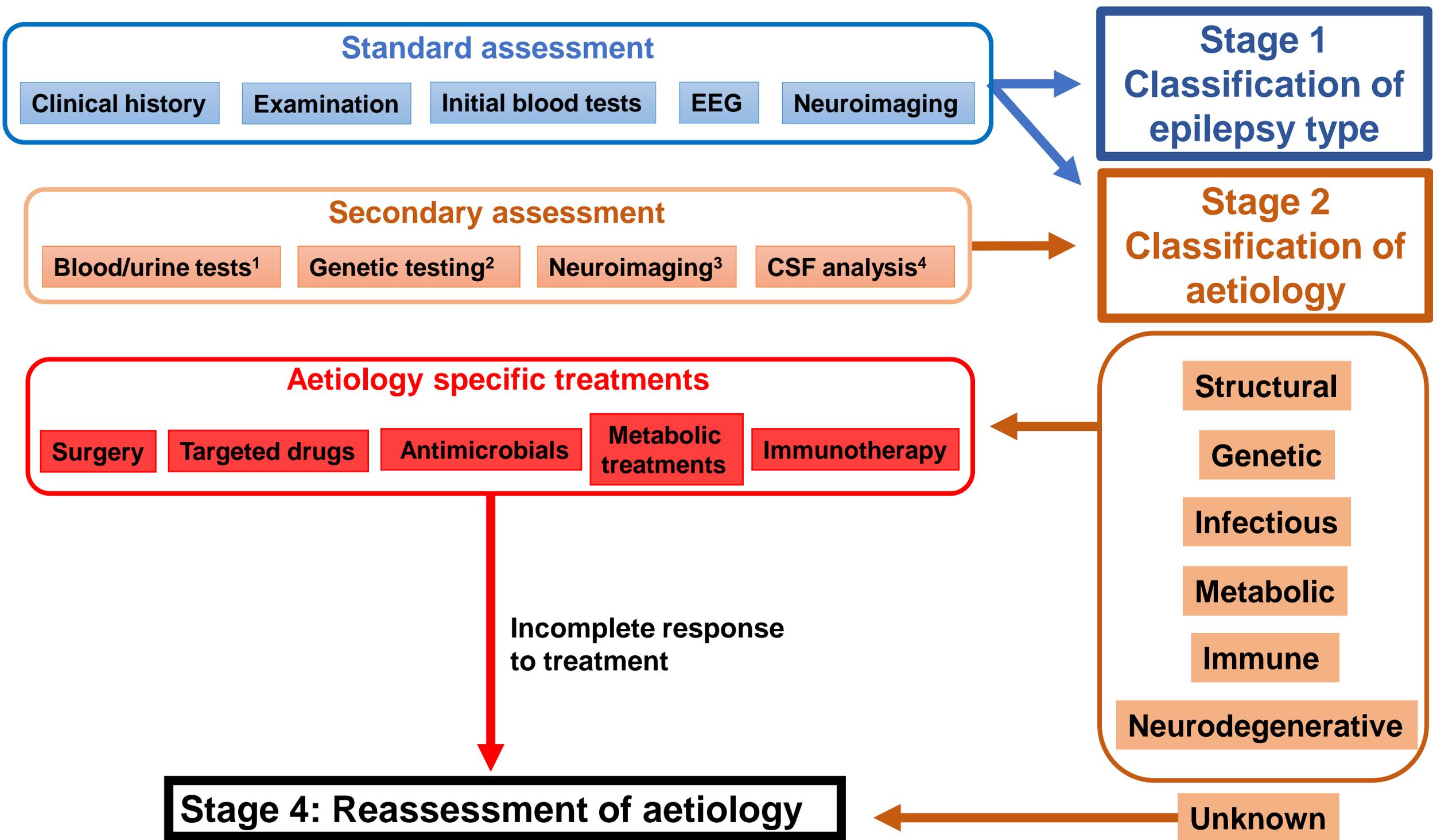
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Key points

- Epilepsy aetiological categories include structural, genetic, infectious, metabolic, immune, neurodegenerative and unknown.
- Epilepsy aetiological categories are not mutually exclusive.
- Identification of epilepsy aetiology may have relevant implications for clinical management, treatment, and prognostic information.
- The main categories of structural aetiology include: hippocampal sclerosis, brain tumours, malformations of cortical development, vascular malformations, glial scarring and brain inflammation.
- The majority of epilepsies have a polygenic contribution, with multiple gene mutations, each with a weak effect size. Therefore, genetic testing often fails to reveal clear genetic causes for common polygenic epilepsies.

Key points

- Infectious causes of seizures often constitute medical emergencies and prompt treatment should be directed both at the infectious agent and at the seizures; seizures are an independent risk factor for mortality in patients with cerebral infections
- Inborn errors of metabolism are commonly associated with seizures and epilepsy, especially in neonates and young children and often the epilepsy responds well to the correction of the metabolic deficit, rather than to antiseizure medication.
- Whilst seizures in the context of autoimmune encephalitis associated with surface-directed antibodies respond well to immunotherapy and do not tend to result in chronic epilepsy, seizures associated with antibodies directed against intracellular antibodies respond poorly to immunosuppression and commonly progress to chronic epilepsy.
- In patients with neurodegenerative disorders, structural, genetic, metabolic, and/or other aetiological factors may contribute to the pathophysiology of epilepsy.



¹Inflammatory indices, autoantibodies, metabolic screening

²chromosomal microarray, single gene testing, gene panel, whole exome/genome sequencing

³Advanced MRI analysis, PET, SPECT

⁴inflammatory indices, isolation of infectious agents, neuronal autoantibodies