



AMBULATÓRIO DE DISTÚRBIOS
DE MOVIMENTO

HC • UFMG

Approach to Chorea

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Disclosures

- None

Summary

- Classification of Chorea
- Chorea in Children
- Chorea in Adults
- Conclusions

Classification of Choreas

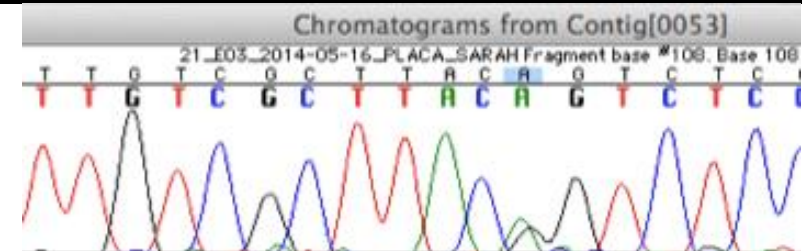
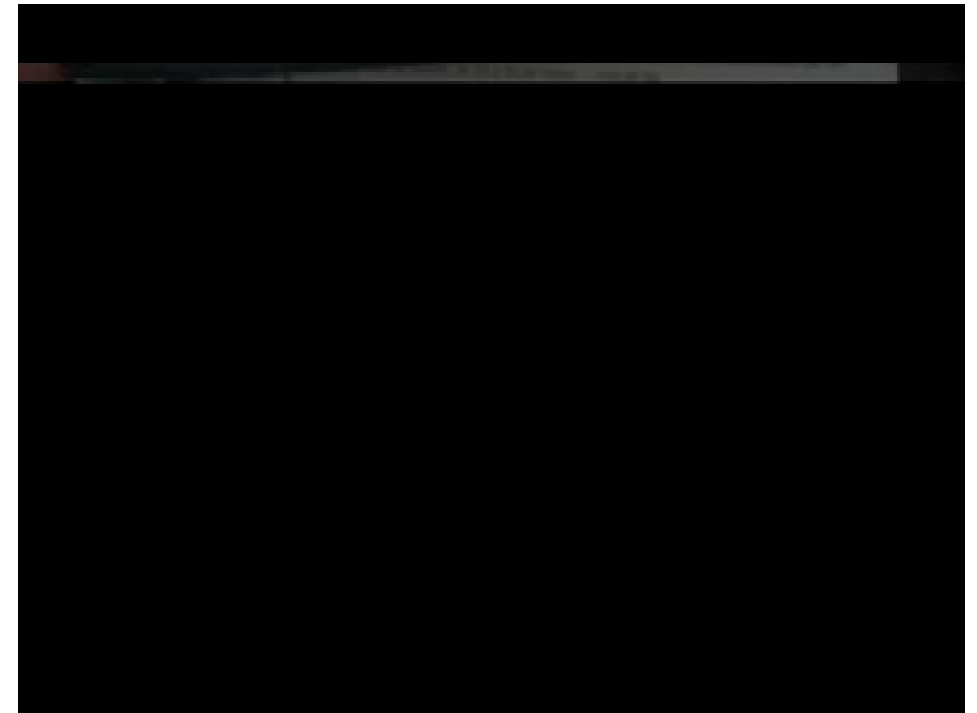
- Genetic Choreas
- Non-Genetic Choreas
 - Drug-Induced Chorea
 - Vascular Chorea
 - Infectious Chorea
 - Immune Chorea
 - Endocrine-Metabolic Chorea
 - Miscellaneous Choreas

Chorea in Children

- Genetic chorea
 - Benign Hereditary Chorea
 - ADCY5 Mutations
 - Many others...
- Acquired chorea
 - Sydenham chorea– 79/82
 - Basal ganglia stroke – 2/82
 - Post-op ischaemic brain changes – 1/82
 - Psychogenic/functional forms

Benign Hereditary Chorea

- Autosomal dominant
- NKX2.1(TITF1) gene (Ch 14q)
- Childhood onset
- Chorea
- (Cognitive problem)
- (Psychosis)
- Hypothyroidism
- Lung disease



ADCY5 Mutations



- Ch 3q21.1
- Gene ADCY5
- Autosomal dominant
- Phenotype
 - Chorea
 - Other dyskinesias
 - Spasticity
 - Cognitive slowness

Sydenham Chorea

- The most common cause of acute chorea in children worldwide
- Age at onset: 9.2 ± 2.7 years
- 25-15% of acute rheumatic fever
- More common in girls (M/F 0.86)
- Carditis in up to 80% of patients
- Arthritis in less than 1/3 of cases
- Behavioral and cognitive features



Sydenham Chorea

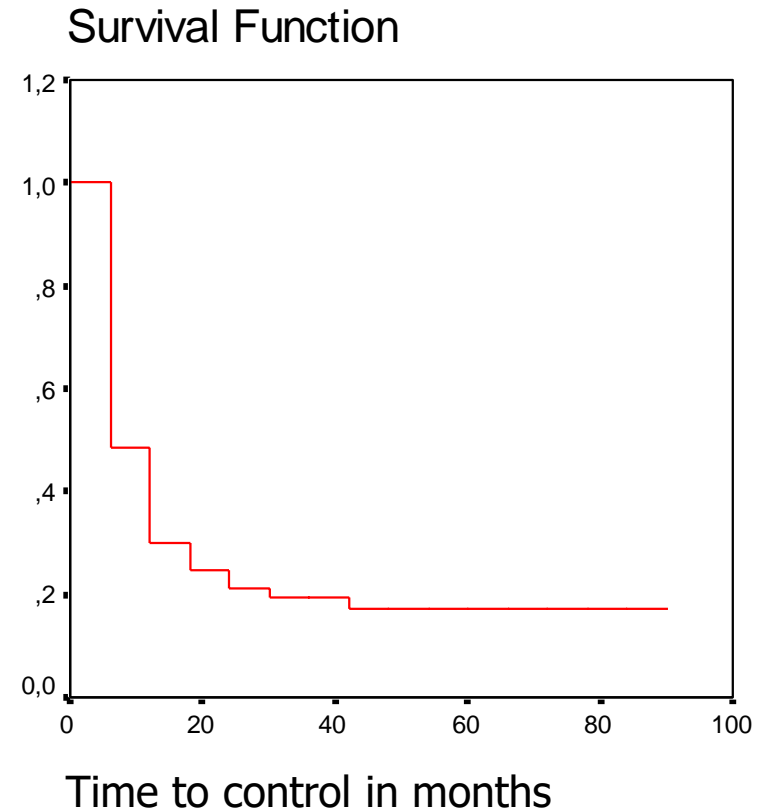
■ Management

■ Chorea

- Valproic acid
- Neuroleptics
- Steroid


■ Secondary Prophylaxis

- Penicillin
- Sulpha





Chorea in children: etiology, diagnostic approach and management

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Received: 30 April 2020 / Accepted: 1 August 2020
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Abstract

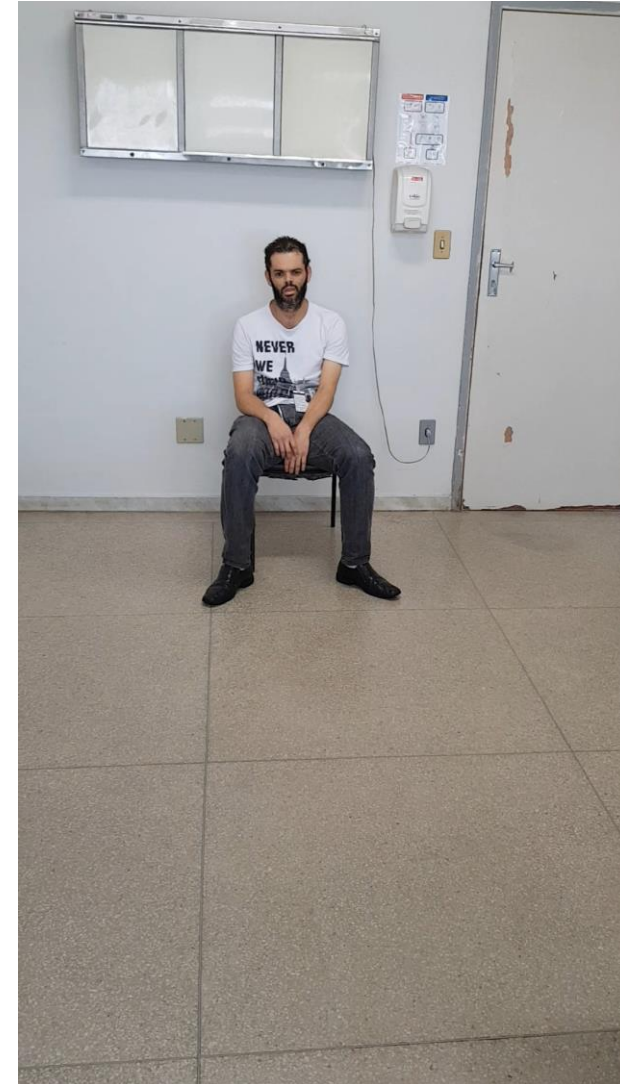
Chorea is defined by the presence of abnormal, involuntary, continuous, random movements that results from a number of autoimmune, hereditary, vascular, metabolic, drug-induced and functional (psychogenic) causes. Chorea may present at all stages of life, from newborns to elderly individuals. While Huntington disease is the main suspicion in adults presenting with chorea, once a drug-induced or parkinsonian dyskinesia have been ruled out; Huntington disease exceptionally presents with chorea in children. Sydenham chorea is considered the most common cause of acute childhood-onset chorea, but its prevalence has decreased in Western countries. However, in younger children other etiologies such as dyskinetic cerebral palsy, anti-NMDAR receptor encephalitis, other autoimmune conditions, or mutations in NKX2-1, ADCY-5, FOXP1, GNAO1, GPR88, SLC2A1, SQSTM1, ATP8A2, or SYT-1 should be considered. In this manuscript, we review the main causes, diagnosis and management of chorea in children.

Chorea in Adults

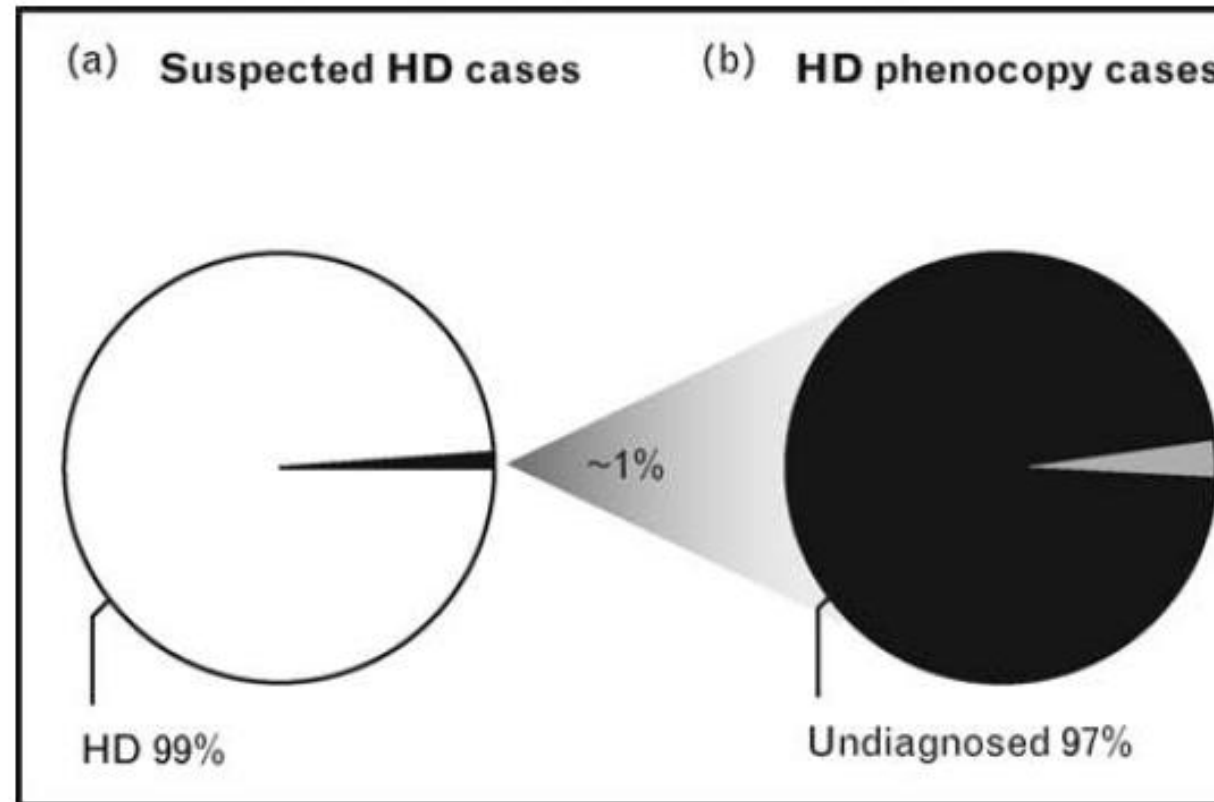
- Genetic Choreas
 - Huntington Disease
 - Huntington Phenocopies (Huntington Disease-Like)
 - Benign Hereditary Chorea
 - Many, many others
- Non-Genetic Choreas
 - Stroke – 21/42
 - Drugs – 7/42
 - AIDS – 5/42
 - Hyperglycemia – 2/42
 - Hyponatremia – 2/42
 - Hypoxia – 2/42
 - Borreliosis – 1/42
 - Vasculitis – 1/42
 - Sydenham chorea – 1/42

Huntington Disease

- Autosomal dominant
- Gene IT15 (Ch4)
- Full penetrance if CAG >38
- Anticipation
- Clinical triad
 - Movement disorders
 - Dementia
 - Behavioral changes



HD Phenocopies



Huntington Disease Like Type 2



C9orf72

- Hexanucleotide (GGGGCC) expansion
- The most common genetic cause of ALS-FTD
- Autosomal dominant
- Up to 1/3 may lack family history
- Variable phenotype
 - Parkinsonism, tremor, and myoclonus
 - Chorea

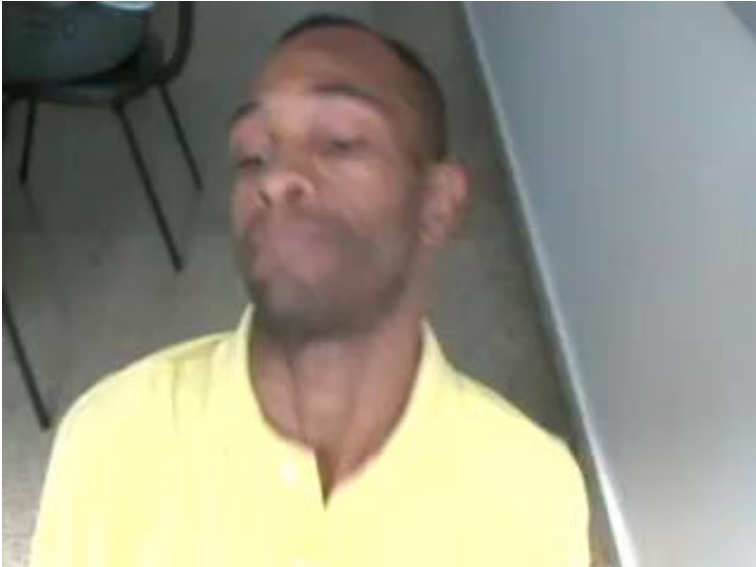
DRPLA

- Dentatorubral-pallidoluysian atrophy
- Autosomal dominant
- >48 CAG repeats on Ch 12p13.31
- 0.48/100.000 in Japan
- 0.33/100.000 in Portugal (2nd only to SCA3)
- Rare cases in Brazil (1/700 patients)
- Dementia, chorea, ataxia and white matter changes on MRI in adult-onset cases
- Myoclonic epilepsy, dementia and cerebral atrophy in early-onset cases

DRPLA

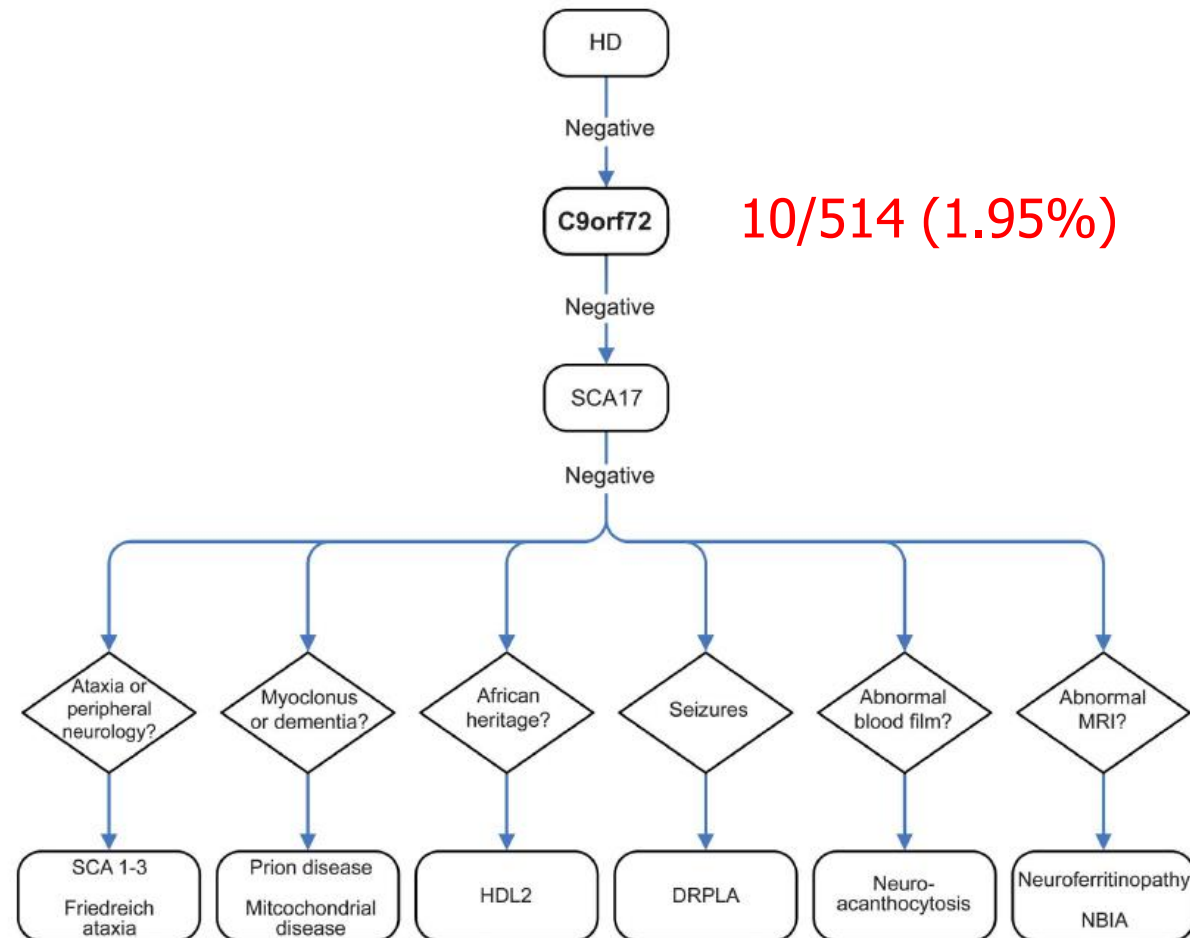
Case 1

Chorea-Acanthocytosis

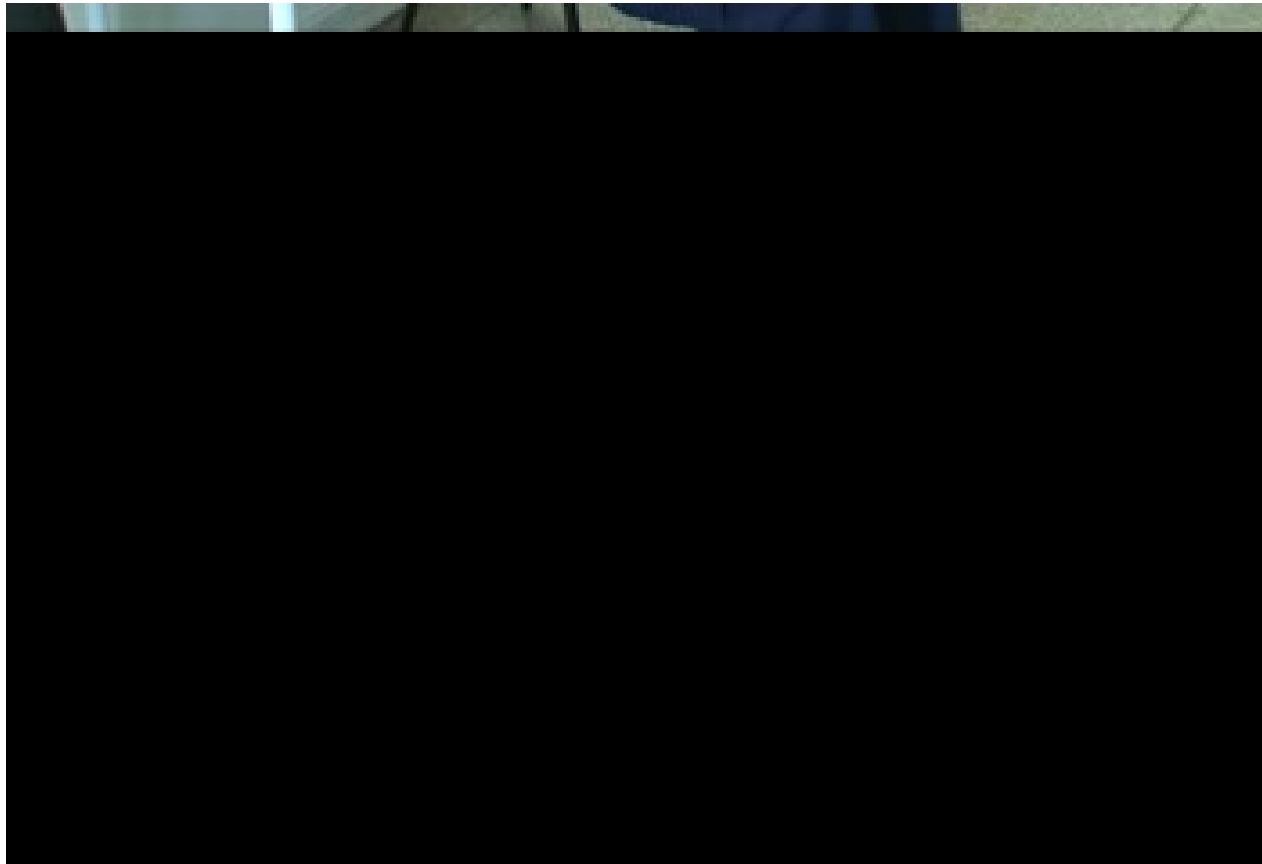


- Autosomal recessive
- Seizure disorder
- Peripheral neuropathy
- Orolingual dyskinesia
- Cognition relatively preserved
- >10% acanthocytes
- Caudate head atrophy

HD Phenocopies in the UK



Drug-Induced Chorea

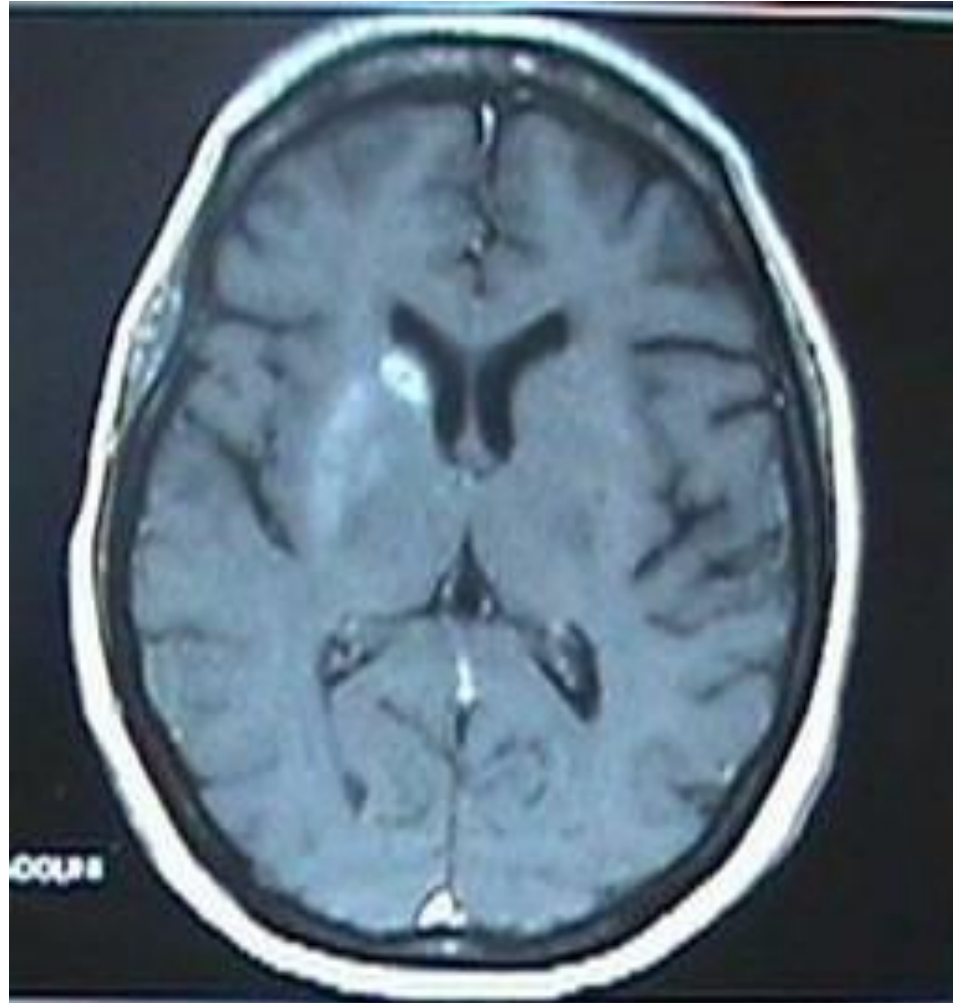


Hemichorea-Hemiballism

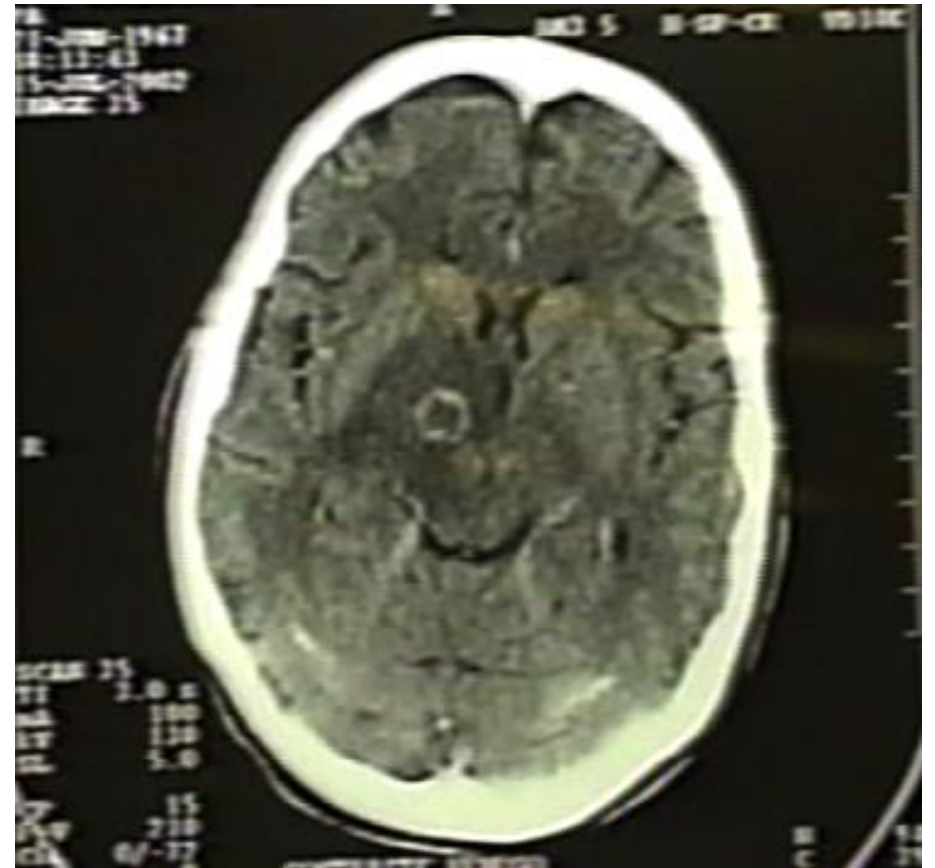


- Etiology
 - Stroke
 - Others are rare
- Management
 - D2R blockers
 - VMAT2 inhibitors
 - Surgery

Chorea and Hyperosmolar Hyperglycemia



HIV and Hemichorea-Hemiballism

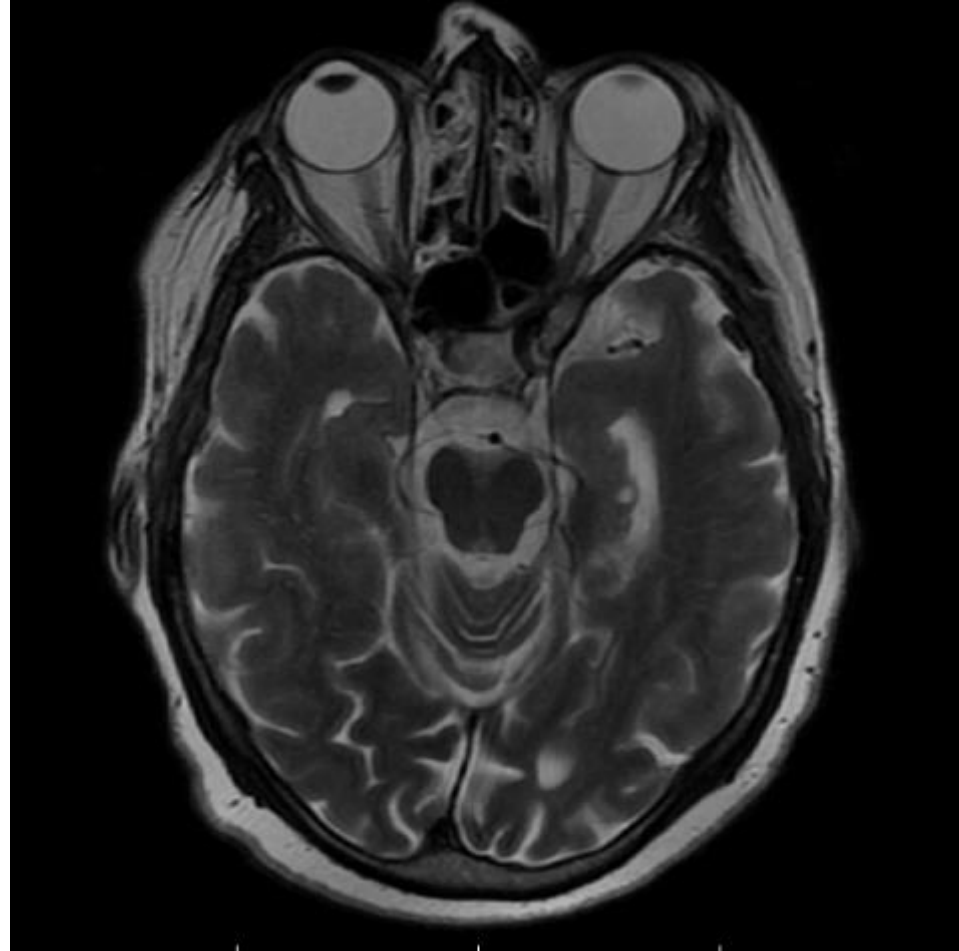


Chorea and SLE

- Neuropsychiatric lupus in 98%
- Chorea and others
- Cognitive and behavioral problems
- Management
 - Symptomatic measures
 - Immune-therapy
 - Anticoagulants



Paraneoplastic Chorea



Anti-CRMP5/CV2 +

Conclusions

- Chorea is characterized by random fragments of motor program
- There are myriad causes of chorea
- The etiology of chorea depends on the age of onset
- Sydenham chorea is the most common acquired chorea in children
- Benign hereditary chorea is the most common genetic chorea in children
- Huntington disease is the most common genetic chorea in adults
- Stroke is the most common acquired chorea in adults



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