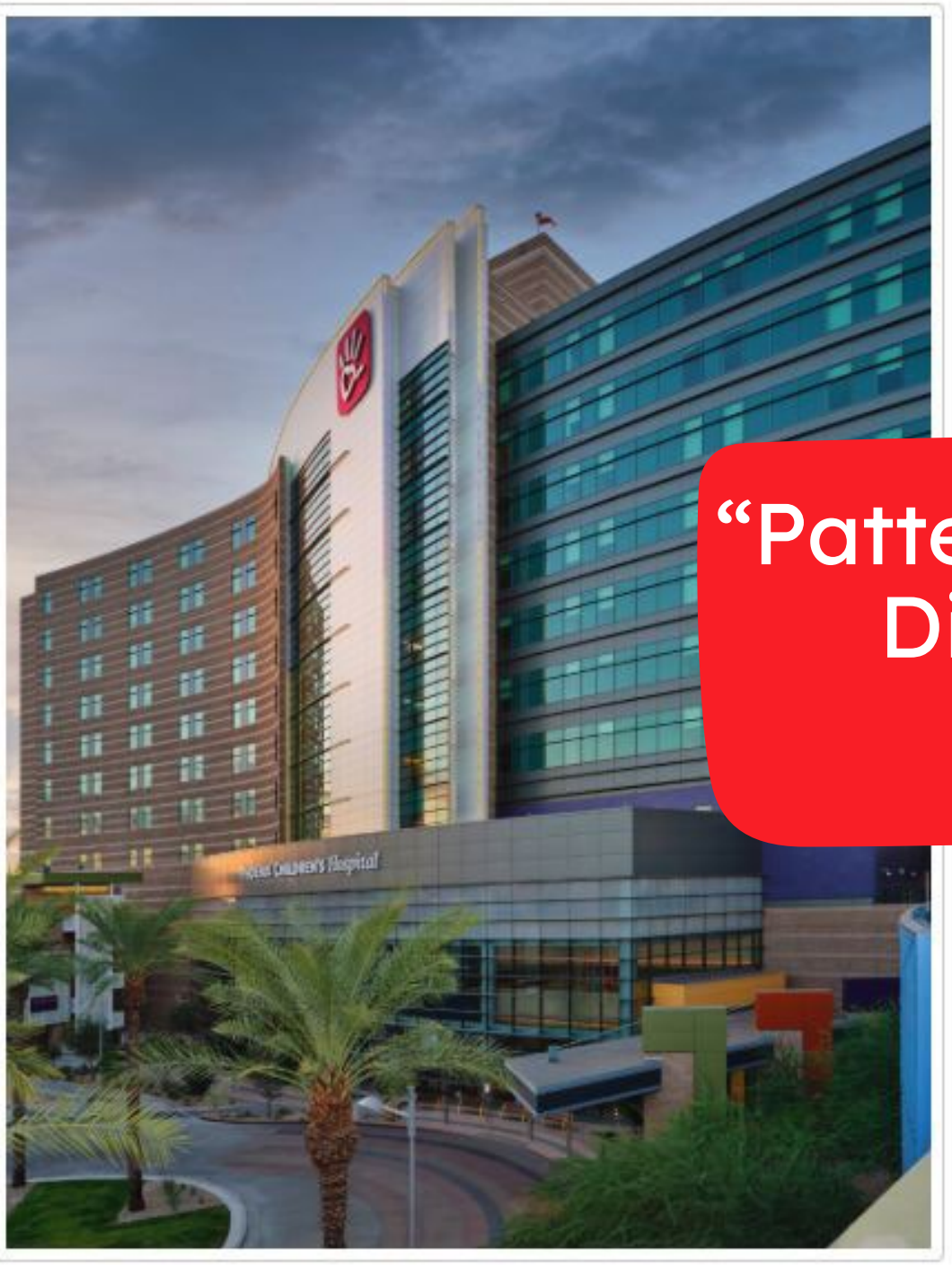

Phoenix Children's

"Pattern Recognition in Movement Disorders: Clinical Insights Through Cases"

Pediatric Update Conference 2026

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Disclosures

- No disclosures or conflict of interest

Objectives

- Discuss the diagnostic value of home videos provided by caregivers in evaluating abnormal movements in children
- Differentiate between normal and pathological movement disorders in children utilizing a case-based approach
- Review illustrative video cases to identify "red flag" features that necessitate urgent investigation or specialist referral

Case 1

There’s a Whole Lot of Shaking Going On ……

Diagnosis?

Based on your concerns which of the following is the most appropriate action:

Diagnosis:

Infantile self gratification (masturbation) syndrome

Differential Diagnosis

- Masturbatory behavior has been mistaken for:
  - Epilepsy
  - Abdominal pain,
  - Paroxysmal dystonia or dyskinesia

Infantile Self Gratification Syndrome

- Masturbation or self stimulation of genitalia is a common human behavior and is believed to occur in 90 - 94% of males and 50 – 60% of females at some point during their lifetime[1]
- The concept of infantile masturbation (IM) was suggested by Still in 1909 and has been widely recognized since then by the medical fraternity[2]
- Masturbatory activity in infants and young children is difficult to recognize because it often does not involve manual stimulation of the genitalia at all
- Parents prefer the term self gratification (or even benign idiopathic infantile dyskinesia)

[1]Leung AK, Robson WL. Clin Pediatr 1993;32:238–41
[2]Still GF. Common Disorders and Diseases of Childhood. London, United Kingdom: Oxford University Press; 1909:336–380

Infantile Self Gratification Syndrome

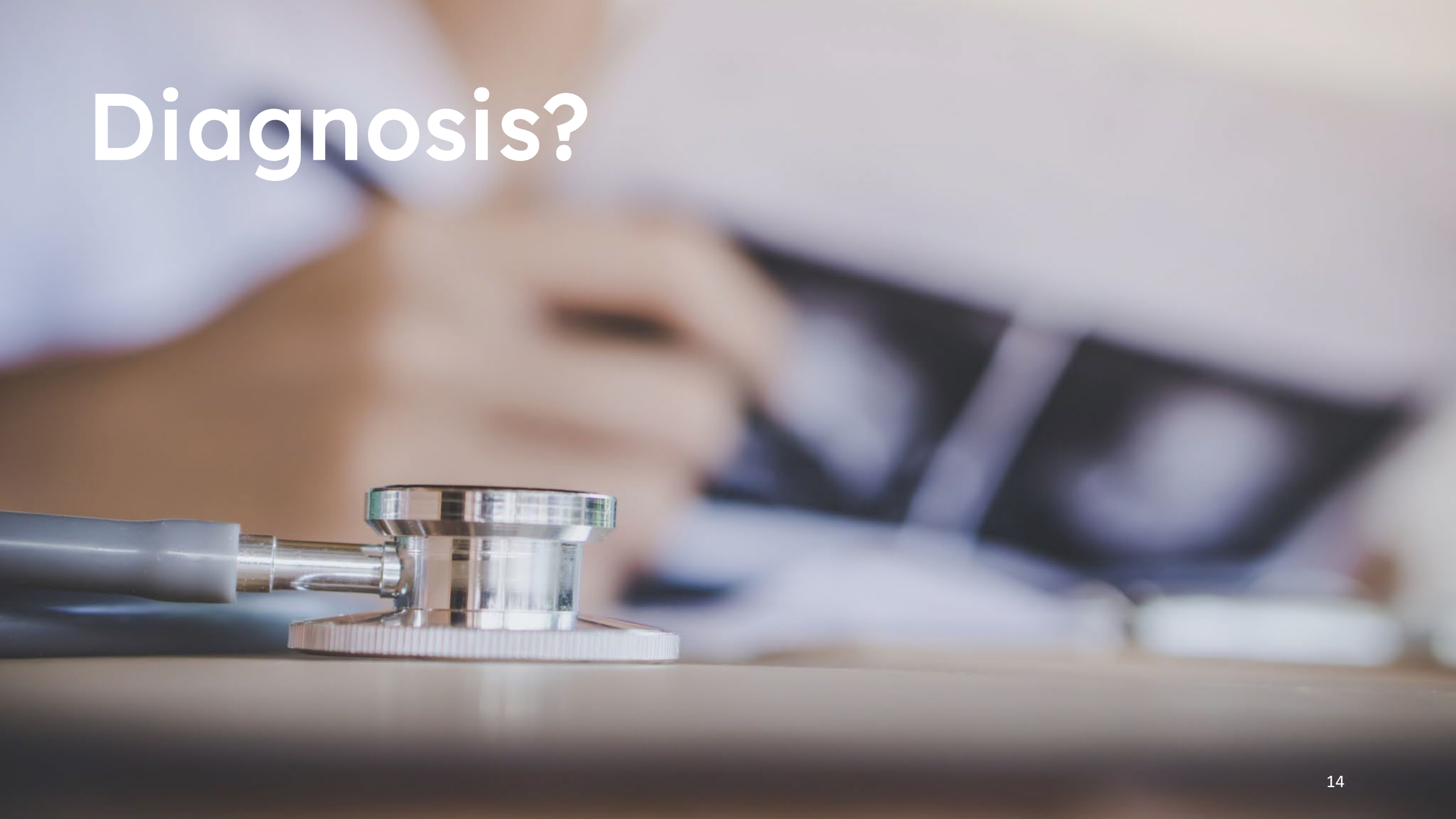
- Infantile self gratification (masturbation) syndrome usually starts by 2 months of age and progressively increases and peaks at 4 years of age
- Secondary peaking occurs during adolescence in both sexes
- The frequency of events varies from 1/week to 12/day (mean: 16/week) and the duration ranges from 30 seconds to 2 hours (mean: 9 minutes)

Yang ML, et al. Pediatrics 2005;116:1427–1432

Case 2

Whazz up ……

Diagnosis?



Based on your concerns which of the following is the most appropriate action:

Diagnosis

Benign Shudder Attack

Benign Shudder Attack

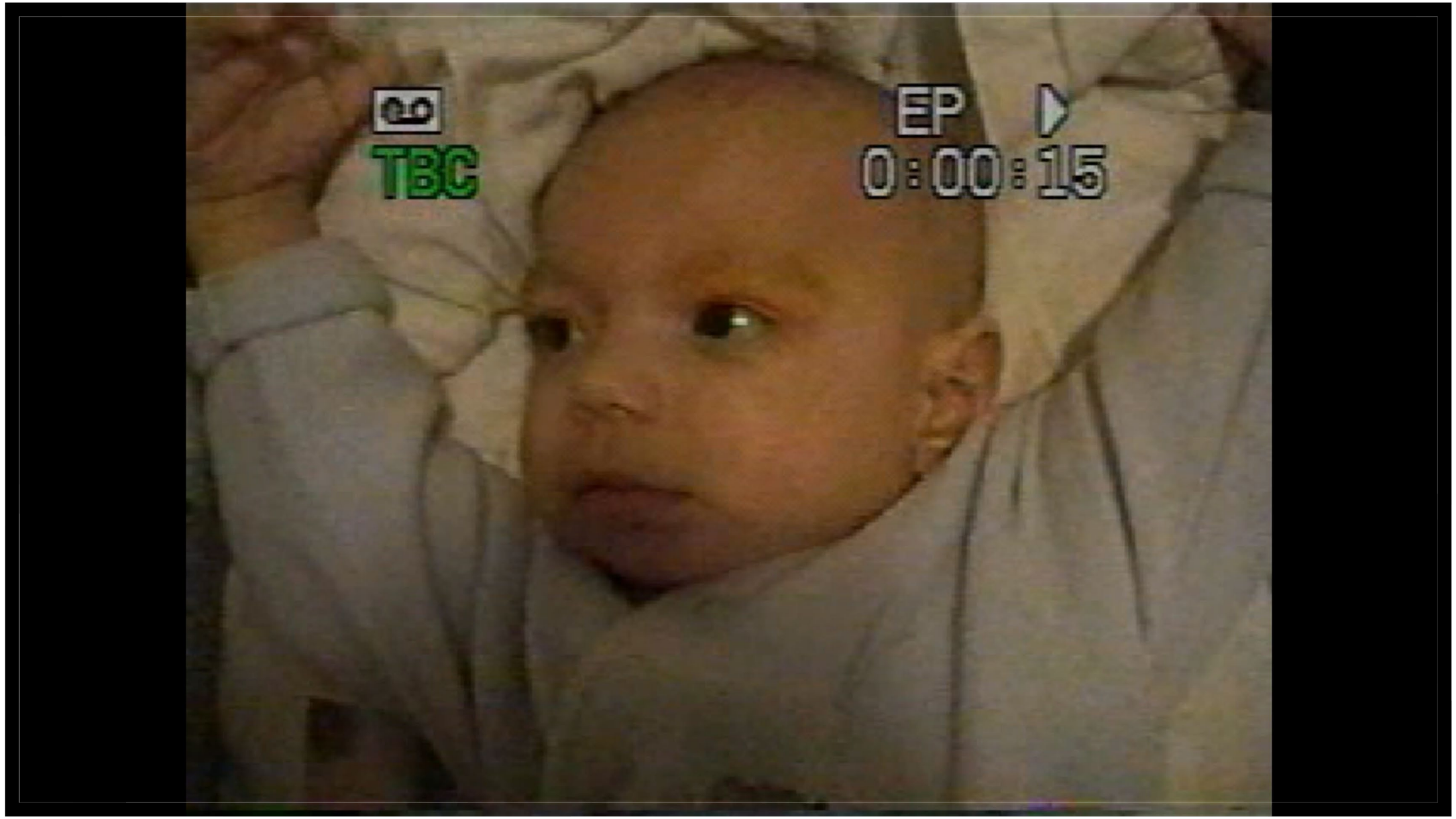
- Start as early as 4 to 6 months of age and rarely occur after the age of 3
- Precipitated or aggravated by excitement, fear, anger, frustration, or embarrassment
- Episodes last usually for a few seconds and are characterized by rapid shivering or stiffening of the body with abnormal posturing with adduction of the knees and arms, flexion of the head, elbows, trunk and knees, and flexion or extension of the neck
- No alteration of consciousness with the abnormal movements

Benign Shudder Attack

- Attacks are very frequent, occurring multiple times a day, and in excess of 100 per day in some cases
- Pathophysiology of shuddering attacks is unknown
- Electromyography studies have revealed the frequency of these shuddering attacks to be similar to that of essential tremor
- Head tremor may evolve from shuddering attacks

Case 3

Oops!...I Did It Again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Diagnosis?

Based on your concerns, this requires urgent attention, evaluation, and treatment to maximize outcome:

Diagnosis

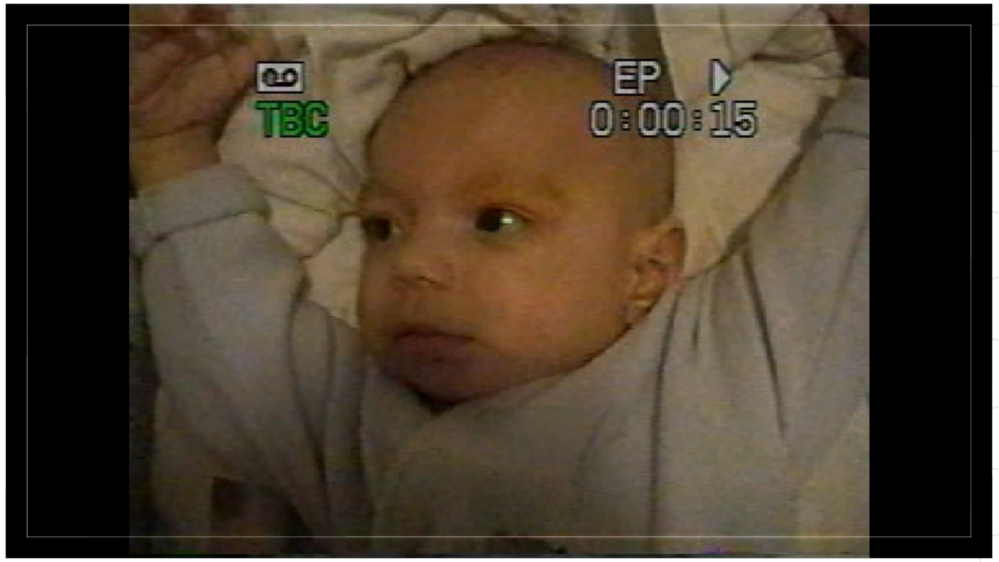
Benign Sleep Myoclonus

Differential Diagnosis: Neonatal/Infantile myoclonus

- Myoclonic epilepsy
  - Benign
  - Progressive myoclonic epilepsies (PME)
- Benign neonatal/infantile sleep myoclonus
- Hyperekplexia
- Myoclonic tics
- Opsoclonus-myoclonus syndrome
- Spinal myoclonus
- Other

Diagnosis: Sleep myoclonus

- Myoclonus is part of normal sleep physiology, as paradoxical excitation in rapid-eye-movement sleep
- Beginning in fetal life, this is most abundant during the first 6 to 8 months postnatally and persists through life as fragmentary nocturnal myoclonus
- Hypnic jerks, or myoclonus on sleep initiation, are associated with the sensation of falling



Myoclonus

- Brief involuntary muscle jerk originating in the central nervous system[1]
  - Cortical – tends to be focal and distal and typically found in the arm.
  - Subcortical – tends to be both proximal and distal generalized myoclonus, involving both agonist and antagonist muscle groups
  - Spinal – tends to be limited to muscles innervated by a few or multiple spinal segments and affects predominantly flexor muscles

[1]Pranzatelli M. Seminars in Pediatric Neurology, 2003;10:41-51

Myoclonus Continued

- It is a paroxysmal event that may appear as an isolated finding or as a symptom of many diseases
- Physiologic myoclonus occurs episodically throughout life as hiccoughs and hypnic (sleep) jerks

Clinically Different Forms of Myoclonus

- Three types:
  - Spontaneous
  - Reflex
  - Movement-induced/action myoclonus (most common)
- Myoclonus is also distinguished from other movement disorders by its unusual association with epilepsy and ataxia

Case 4

In my baby’s eyes ……

History

- 12-month-old girl with 3-week history of clumsiness, increased falling, ataxia and abnormal eye movements
- Diarrheal illness 1 week prior to onset of symptoms
- Needing support to sit up
- More recent history of jerking of legs, including while asleep
- No recent immunization
- Previously healthy child

Diagnosis?

Based on history and your observation, the most likely diagnosis is:

Diagnosis?

Opsoclonus-myoclonus syndrome

Differential Diagnosis

- Acute cerebellar ataxia
- Postinfectious cerebellar ataxia
- Miller-Fisher variant of Guillain Barre syndrome
- Acute disseminating encephalomyelitis (ADEM)
- Myoclonic epilepsy
- Opsoclonus-myoclonus syndrome

Opsoclonus-myoclonus Syndrome

- Synonyms: Kinsbourne syndrome, Dancing Eyes-Dancing Feet Syndrome, Opsoclonus myoclonus ataxia syndrome
- Characterized by:
  - Opsoclonus
  - Myoclonus
  - Ataxia
  - Encephalopathy

Opsoclonus

- Disorder of ocular motility characterized by spontaneous, arrhythmic (chaotic), conjugate saccades occurring in all directions of gaze without a saccadic interval

Overview

- Autoimmune neurological disorder.
- Onset typically < 2 years of age
- Males and females equally affected
- **Paraneoplastic versus infectious in origin**
- Myoclonus is maximal when movement is attempted but may be present at rest
  - Worsens with agitation or stimulation
  - Child may appear tremulous (polyminimyoclonus) or have gross jerking
  - Face, eyelids, limbs, fingers, head and trunk are involved

Common Associated Features

- Irritability
- Reduced sleep
- Dysarthria or mutism
- Hypotonia

Tumor Association

- Tumors are found in about half the cases of opsoclonus-myoclonus
- Most common tumors are neuroblastoma and ganglioneuroblastoma which occur often in the chest, but also in the abdomen or pelvis.
- These tumors derive from neural crest cells
- Affects 2 to 3% of children with neuroblastoma
- Molecular mimicry/paraneoplastic
  - Involves brain regions like the cerebellum, brainstem, and limbic system

Prognosis

- Often a chronic-relapsing and debilitating illness
- Severity of the initial symptoms may predict a chronic-relapsing disease course and learning disability[1]
  - Tumor status or time interval and response to treatment did not.
- Neurodisability in ~ 60-80% of patients
  - Motor abnormality/ataxia
  - Speech abnormality
  - Learning disability
  - Behavioral problems
  - Persistent opsoclonus (less common)

[1]Brunklaus a et al. Pediatrics 2011;128:e388–e394

Case 5

Things That Go Bump In The Night

History

- 4-year-old boy with abrupt onset rhythmic movement during sleep
- Occurs in clusters – lasting up to 5 minutes
- Normal development
- No past medical history of note
- Normal neurological examination

Diagnosis?

Based on your observations, EEG is likely to be:

Diagnosis?

Jactatio capitis nocturna (head banging)

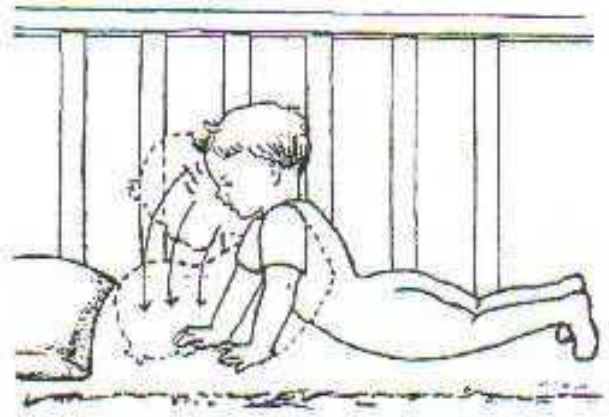
Jactatio capitis nocturna (head banging)

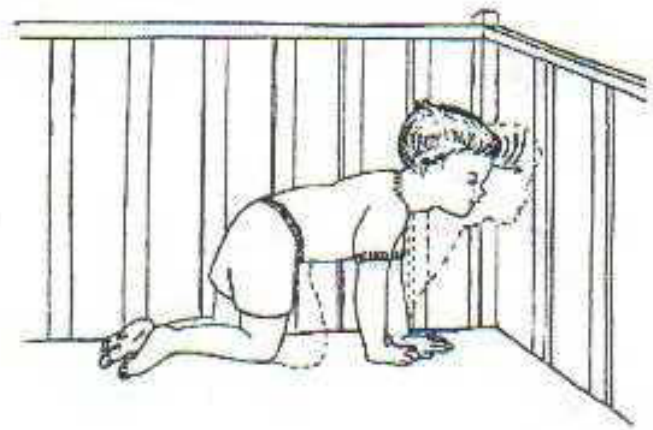
- Head banging is a subtype of sleep related rhythmic movement disorder (RMD)
- It mainly affects infants and children
- Involves large muscle groups (especially neck and trunk muscles) that engage in repetitive, stereotyped, and rhythmic movements

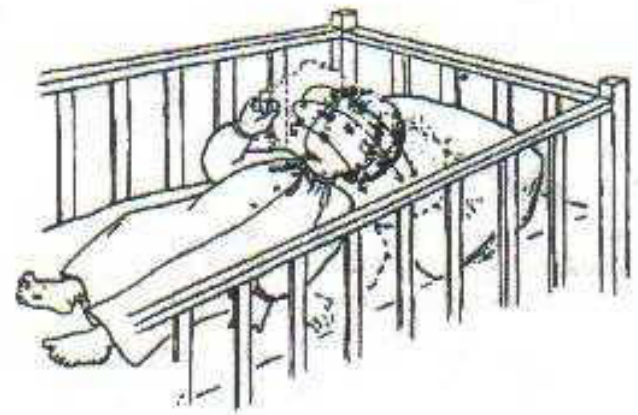
- Movements emerge predominantly during drowsiness or sleep
- Clustered episodes usually lasting less than 15 minute
- Frequency of 0.5-2 Hz

ICSD Diagnostic criteria for RMD

- Rhythmic body movements during drowsiness or sleep (0.5-2Hz).
- At least one of the following types is present:
  - Head- banging
  - Head rolling type
  - Body rocking
  - Body rolling







Typical Movements

- Head banging typically occurs with the child lying face down – banging the head down into a pillow or mattress
- In the upright position, the head is banged against the wall or headboard repeatedly or the upper body may be rocked
- Body rocking is typically done with the entire body while on the hands and knees

Rhythmic Movement Disorders of Sleep

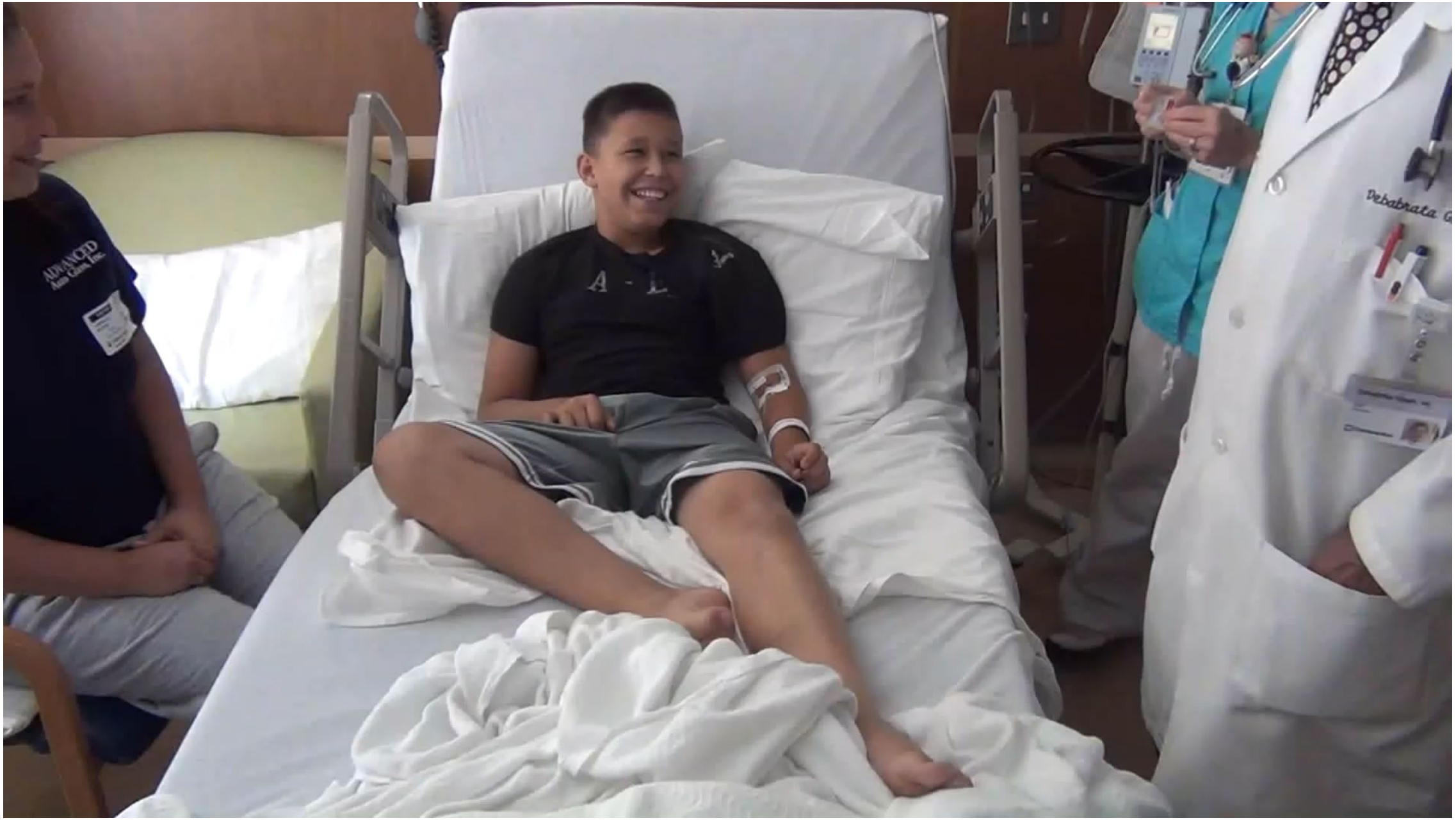
- Usually disappears by age 5
- Continues in 6% of 5-year-olds and in 3% of 13-year-olds
- Benign in the large majority of cases
- Most children and adults who have rhythmic movement disorder are healthy, although the condition is more common in children with autism and other developmental disabilities

TREATMENT

- Typically none
- Medications (rarely)
  - Benzodiazepine (Clonazepam)
  - Antidepressant citalopram
  - Hydroxyzine
  - Antihistamines
  - Gabapentin

Case 6

A Malady Of Movement


ADVANCED

Diagnosis?

Based on your observation, the most likely diagnosis is:

Diagnosis?

Sydenham Chorea

Differential Diagnosis

- Paroxysmal disorders
- Benign hereditary chorea
- Autoimmune
- Infectious/post infectious
- Structural: vascular, neoplastic
- Neurodegenerative
- Drugs
- Pregnancy

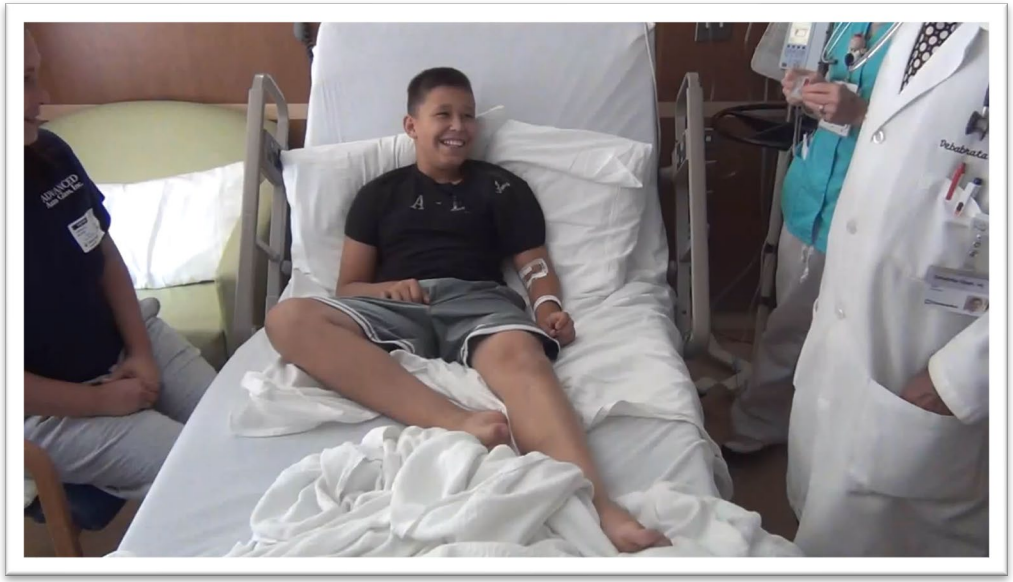
Chorea

- Abnormal involuntary movement derived from the Greek word “dance”.
- Brief, abrupt, irregular, unpredictable, non-stereotyped jerky movements
  - Extremely fidgety, clumsy, uncoordinated and awkward
  - Appear to be “making faces”
- Most often affects the face, arms, and hands

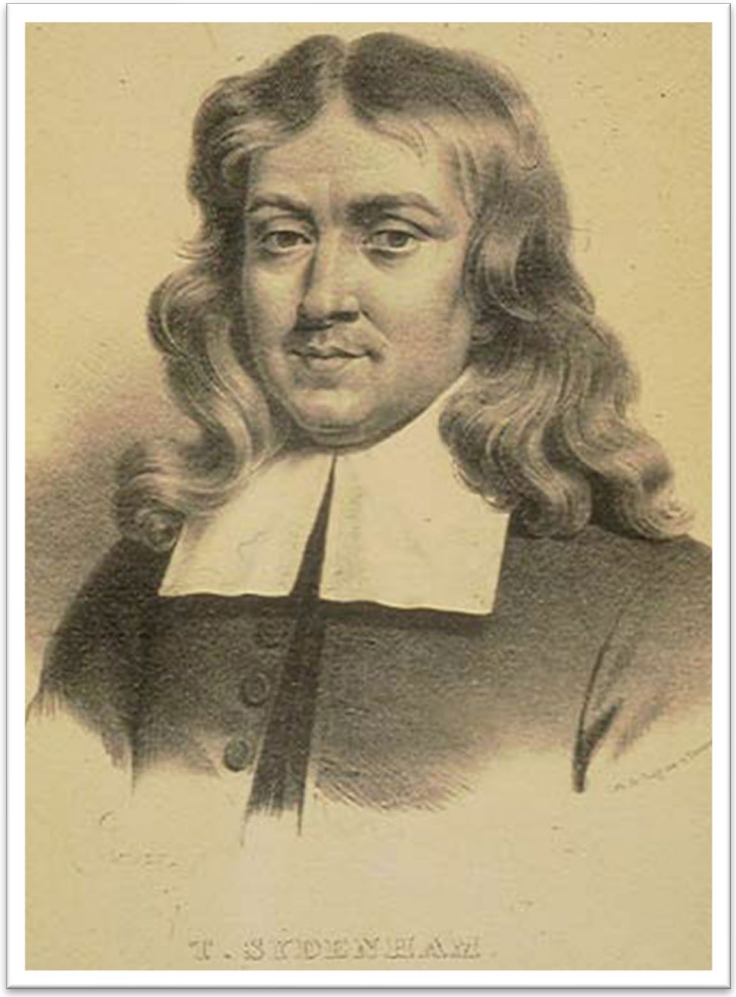
- Abnormal behavioral or emotional problems
- Exacerbated by stress, fatigue or excitement
- May only affect one side of the body
- Treatment: benzodiazepines, valproic acid, pimozide, tetrabenazine, corticosteroids, IVIG, and plasma exchange

Sydenham Chorea

- “St. Vitas Dance”
- Usually develops following Streptococcal infection
- Major complication of acute rheumatic fever
- Considered an autoimmune disorder



- Usual age of affliction is 5 -15 years
- More prevalent in girls than in boys
- Named for the 17th century English physician, Thomas Sydenham



St. Vitus Dance
(Sydenham's Chorea, Infectious Chorea)

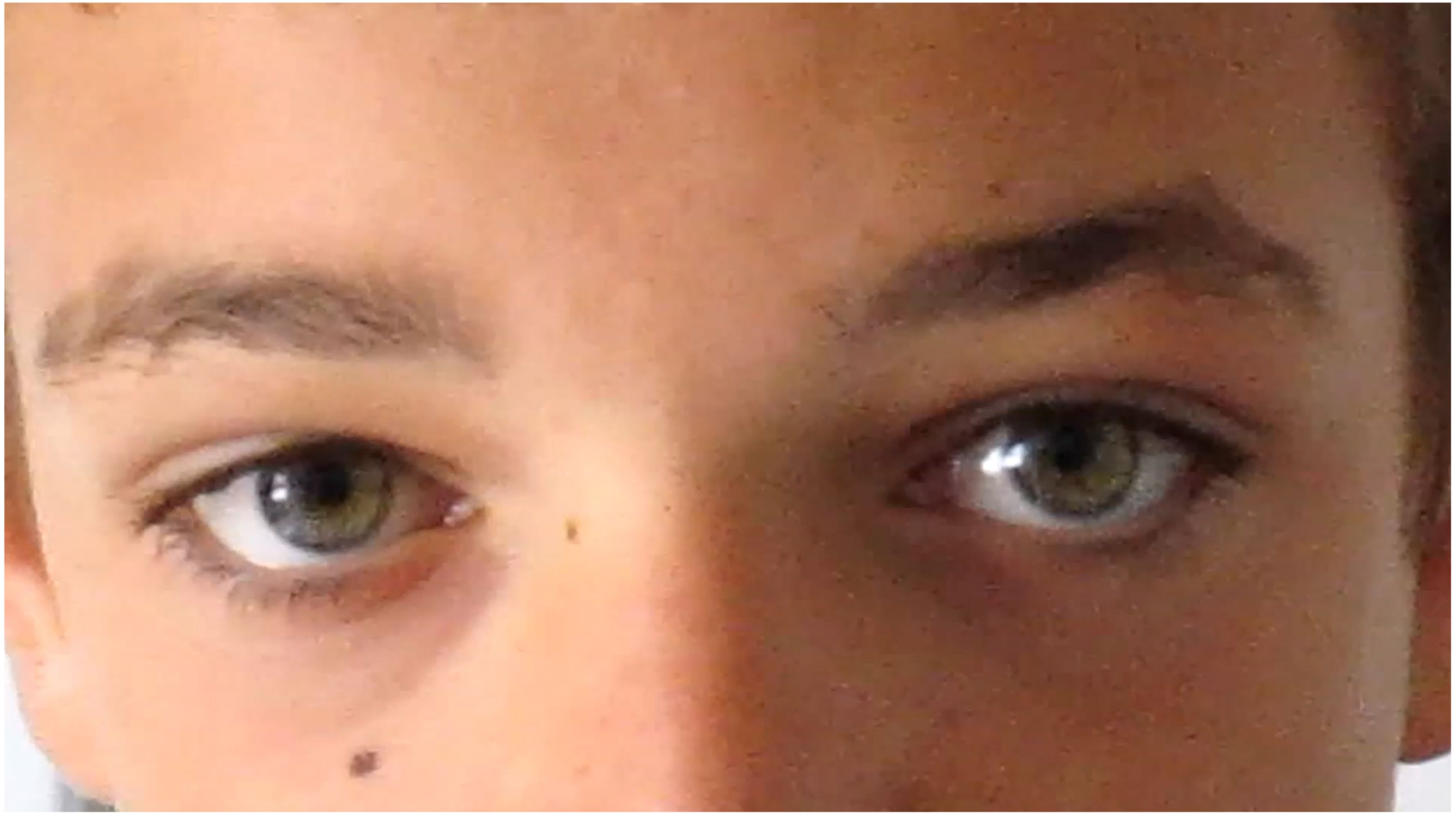
- The medieval name given to Sydenham chorea
- In the late Middle Ages people in Germany celebrated the feast of Vitus by dancing before his statue
- St. Vitus often invoked to alleviate the suffering of people with epilepsy
- Patron saint of dancing, epilepsy and actors

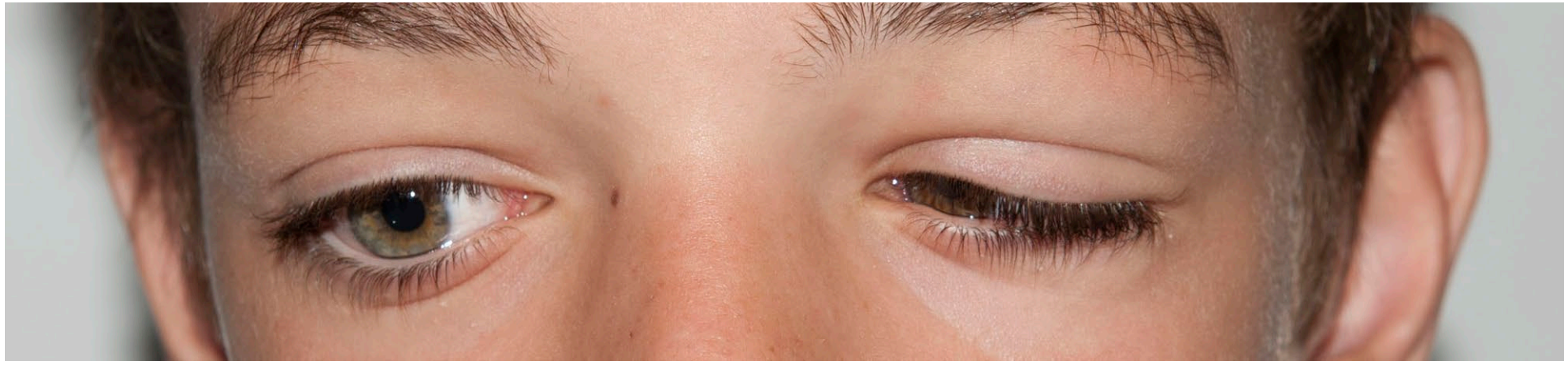
Case 7

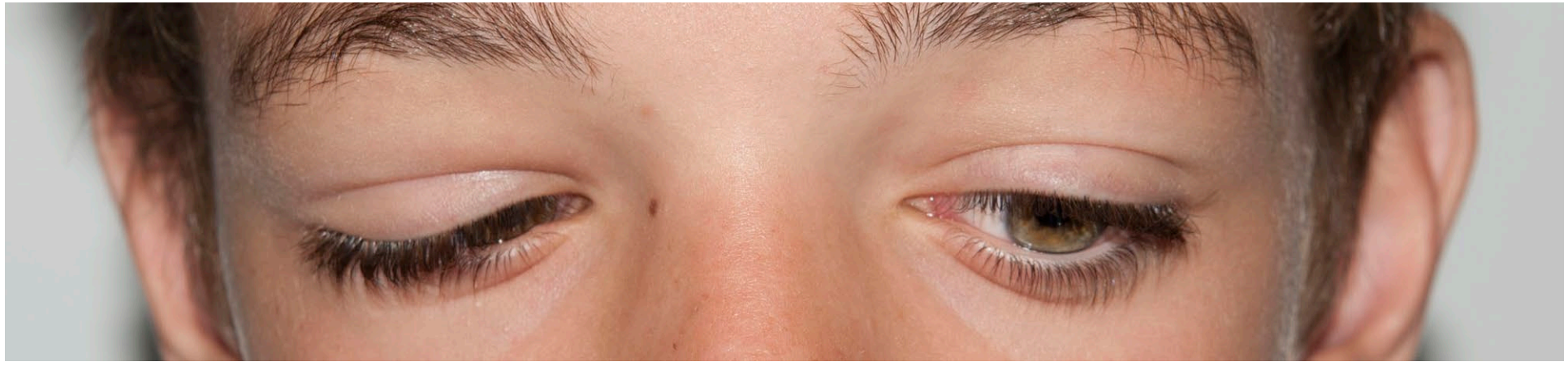
In The Eyes Of The Beholder

History

- 15-year-old boy initially seen for problems of hypotonia as an infant
- Also, problems with early mild developmental delay and some persistent academic and coordination difficulties when older
- Birth and family history non-contributory
- Past medical history otherwise unremarkable







Diagnosis?

Based on your concerns which of the following is the most appropriate action:

Diagnosis?

Duane Syndrome

Duane Syndrome

- A congenital non-progressive eye movement disorder
  - Prevalence 1:1000
  - Slightly more common in females (3:2 ratio)
- Characterized by:
  - Limitation of abduction and/or adduction
  - Globe retraction on adduction of eye
  - Palpebral fissure narrowing on adduction of eye
- Most cases (70%) are isolated with no other congenital anomalies
- Rarely familial – associated with mutations in the *CHN1* gene

Phoenix Children's®

Thank you!

Case 8

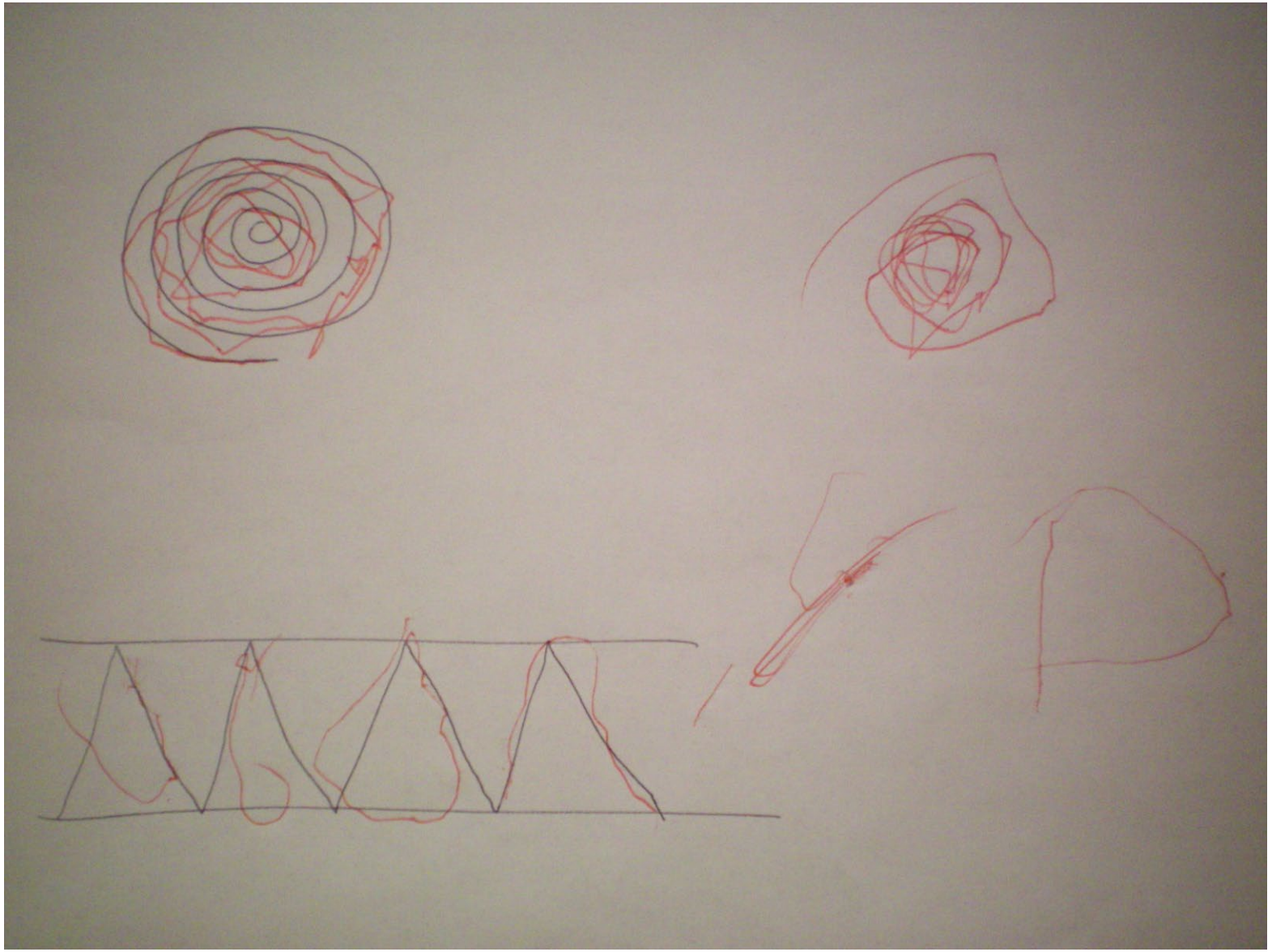
A slipping and sliding ……

Tremors

- Onset 18 - 24 months of age
- Shaking of head and upper extremities during activities
- Exacerbating factors: fine motor activities
- 2 ½ years: started occupational therapy → some improvement

Clumsy Walking

- Onset at 2 years
- Frequent falls while walking
- Exacerbating factors: fatigue, running
- Worse by age 3 years



Investigations

- MRI brain - normal
- MRS brain - normal
- MRI spine - normal
- EMG/NCV - normal
- EEG - normal
- CSF amino acids, IgG index, OCB, neurotransmitters including folate – all normal
- Consults: Rheumatology, ophthalmology, pulmonology – not helpful

Investigations Continued

- CBC, CMP
- Antiphospholipid Ab, Beta 2 glycoprotein IgG and IgM
- Heavy metal screen in urine, Lead level, TFT, HVA, VMA
- Lactate, ammonia, carnitine, acylcarnitine profile, amino acids, VLCFA, urine organic acids
- PT, PTT, LA. Factor V Leiden mutation, prothrombin gene
- C3 level, C4 level, immune complexes, ANA, dsDNA, ESR
- Carbohydrate deficient transferrin
- Vit B12 level, E level, Alpha fetoprotein, Lyme w/u neg, RPR neg
- Serum copper, ceruloplasmin

Pre-treatment

Diagnosis?

Diagnosis

Dystonia Myoclonus Syndrome

Dystonia Myoclonus Syndrome

- Pathogenic mutation in the **SGCE** gene (exon3c.289C>T,p.Arg97X)
- Primary genetic dystonia: DYT11
  - Autosomal dominant; variable penetrance
- Prescribed trihexyphenidyl, but did not tolerate due to mood swings
- Doing well on clonazepam 0.5 mg daily (see video)

Post-treatment

Subgroup	Designation		OMIM	Chromosomal mapping	Disease gene
Pure dystonia	Dystonia 1	Early-onset dystonia, idiopathic torsion dystonia, dystonia musculorum deformans	128 100	9q34	*TOR1A*
	Dystonia 4	Hereditary whispering dysphonia	128 101	–	–
	Dystonia 6	Idiopathic torsion dystonia of 'mixed' type	602 629	8p11.21	*THAP1*
	Dystonia 7	Focal, adult-onset dystonia, idiopathic focal dystonia	602 124	18p	–
	Dystonia 13	Primary dystonia with mixed phenotype	607 671	1p36.13–p36.32	–
Dystonia plus	Dystonia 5a	DRD; Segawa syndrome; hereditary progressive dystonia with marked diurnal fluctuation, HPD	128 230	14q22.1–q22.2	*GCH1*
	Dystonia 11	M-D; alcohol-responsive dystonia	159 900	7q21	*SGCE*
	Dystonia 12	Rapid-onset dystonia-parkinsonism	128 235	19q12–q13.2	*ATP1A3*
	Dystonia 15	M-D	607 488	18p11	–
Paroxysmal dystonia	Dystonia 8	PDC; PNKD1; non-kinesigenic choreoathetosis; Mount-Reback disease	118 800	2q35	*PNKD1/MR1*
	Dystonia 9	PDC with episodic ataxia and spasticity; episodic choreoathetosis/spasticity (CSE)	601 042	1p21–p13.3	–
	Dystonia 10	PKC; paroxysmal familial dystonia; PKD	128 200	16p11.2–q12.1	–
	Dystonia 18	Paroxysmal exertion-induced dyskinesia; paroxysmal exercise-induced dystonia	612 126	1p31.3–p35	*SLC2A1*
	(Dystonia 19)	PKD2	611 031	16q13–q22.1	–
	(Dystonia 20)	PNKD2	611 147	2q31	–

Muller U. The monogenic primary dystonias. Brain. 2009;132:2005-25.

Case 9

Shake, rattle and roll .....

Referred for Tremor

- 15-yearold boy
- Tremor in left fingers for the past one year
- Difficulty in using his fingers while playing piano
- Feeling of “cramp” when he cannot stretch his fingers
- Left hand muscle weakness

History

- Worse when exposed to cold, better with warmth
- Cramping pain and stretching sensation
- Symptoms are spreading to the right fingers
- Progressively worsening

History Continued

- No weakness in lower limbs
- No bladder or bowel function
- No visual or hearing problems
- No problems with swallowing
- No history of seizures or headaches
- No hearing and vision problem
- No history of any febrile or flu-like illness

- Birth, Development, Past Medical and Family History: all unremarkable

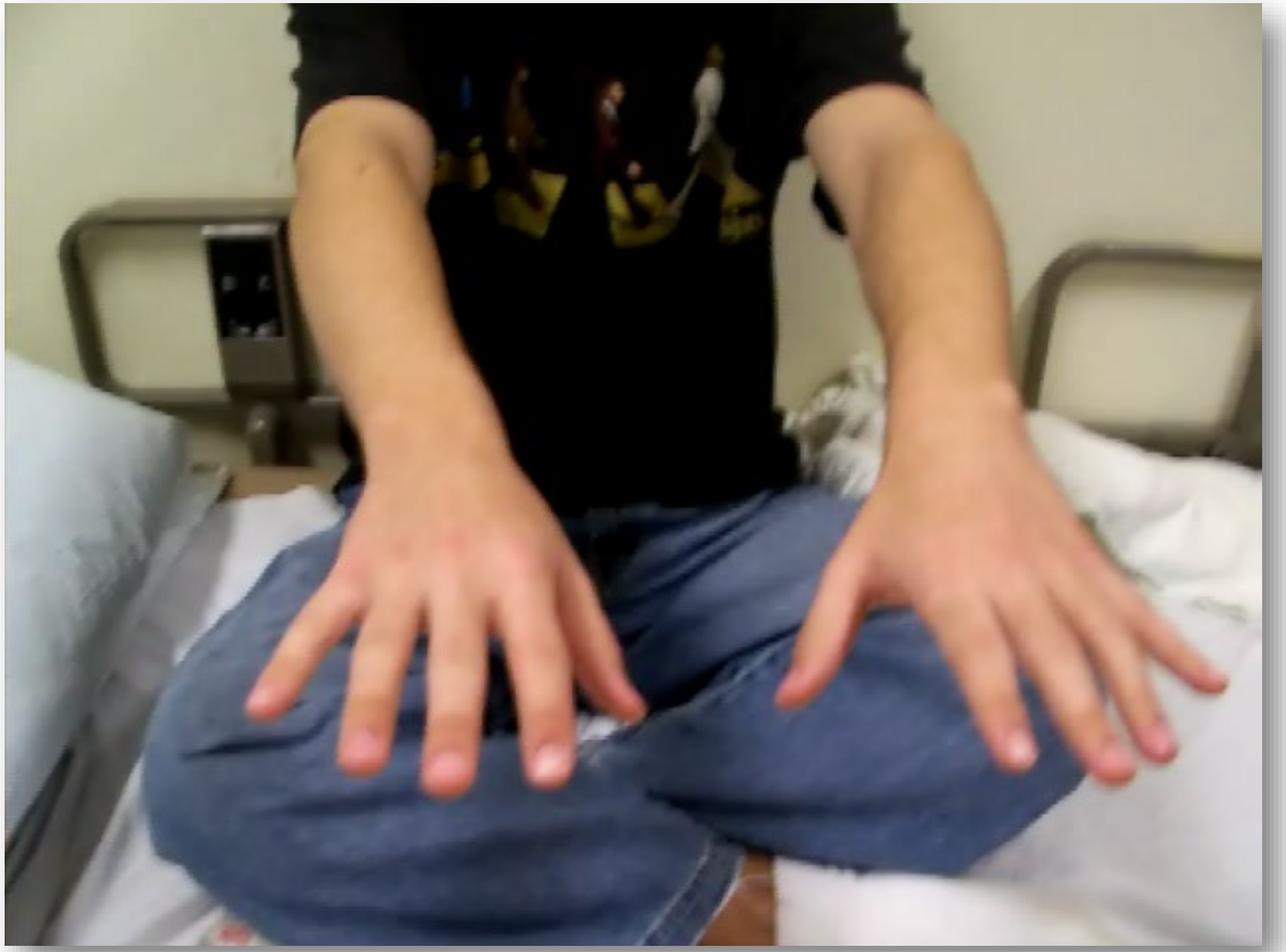
Examination

- Higher function- normal
- Cranial nerves II - XII are intact
- Motor: Normal muscle bulk and tone
  - **Reduction of bulk in left forearm**
  - **Wasting of small muscle of hand left > right**
  - **Mild clawing of left hand**
- Strength otherwise MRC grade 5 in both upper and lower extremities, proximally and distally
- Reflexes 2+/4 symmetrical and equal
- Plantar response is flexor

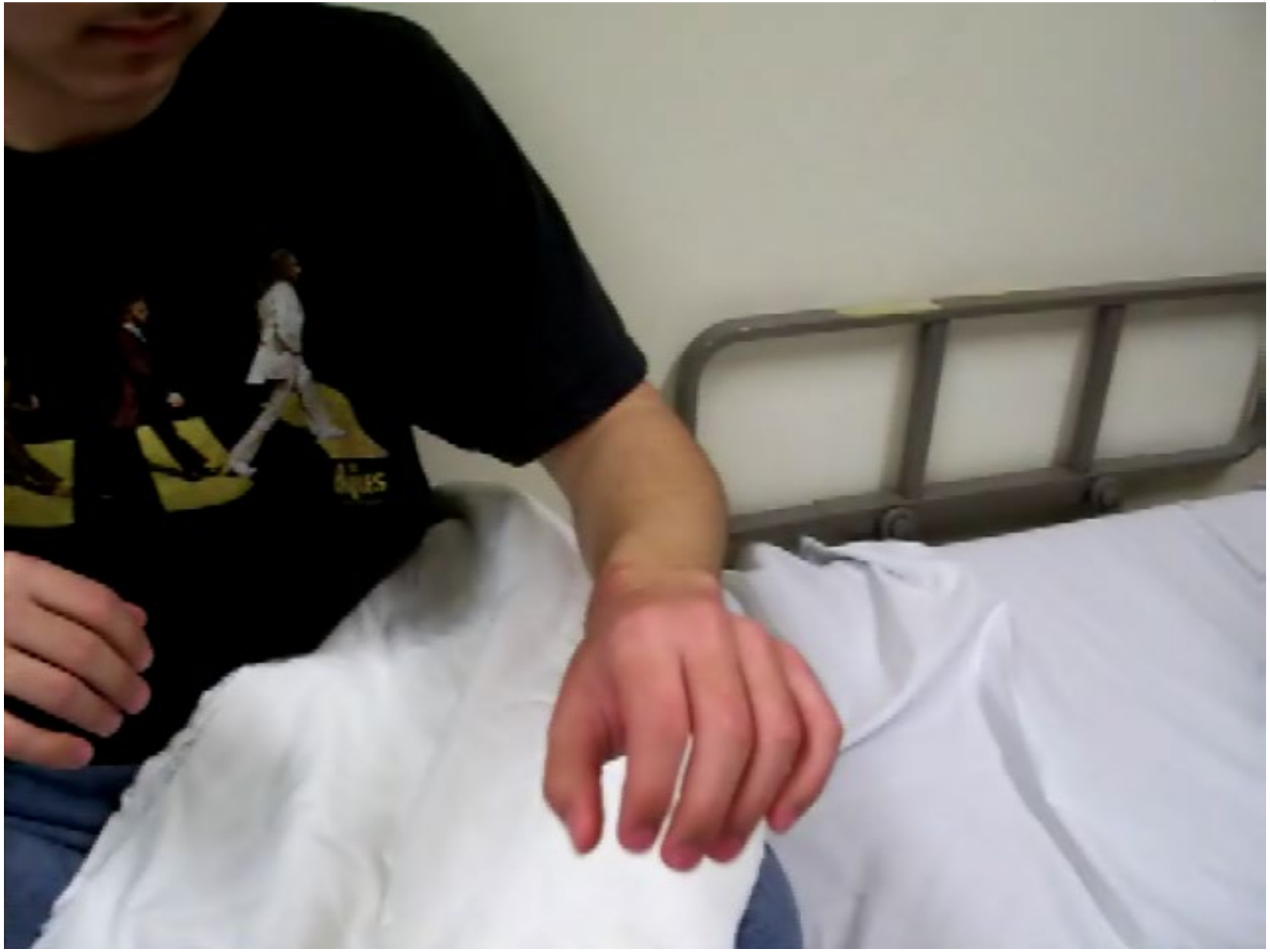
Examination Continued

- Sensory examination completely normal
- Normal cerebellar examination
- Romberg is negative
- Gait is normal
- No other neuroaxis involvement

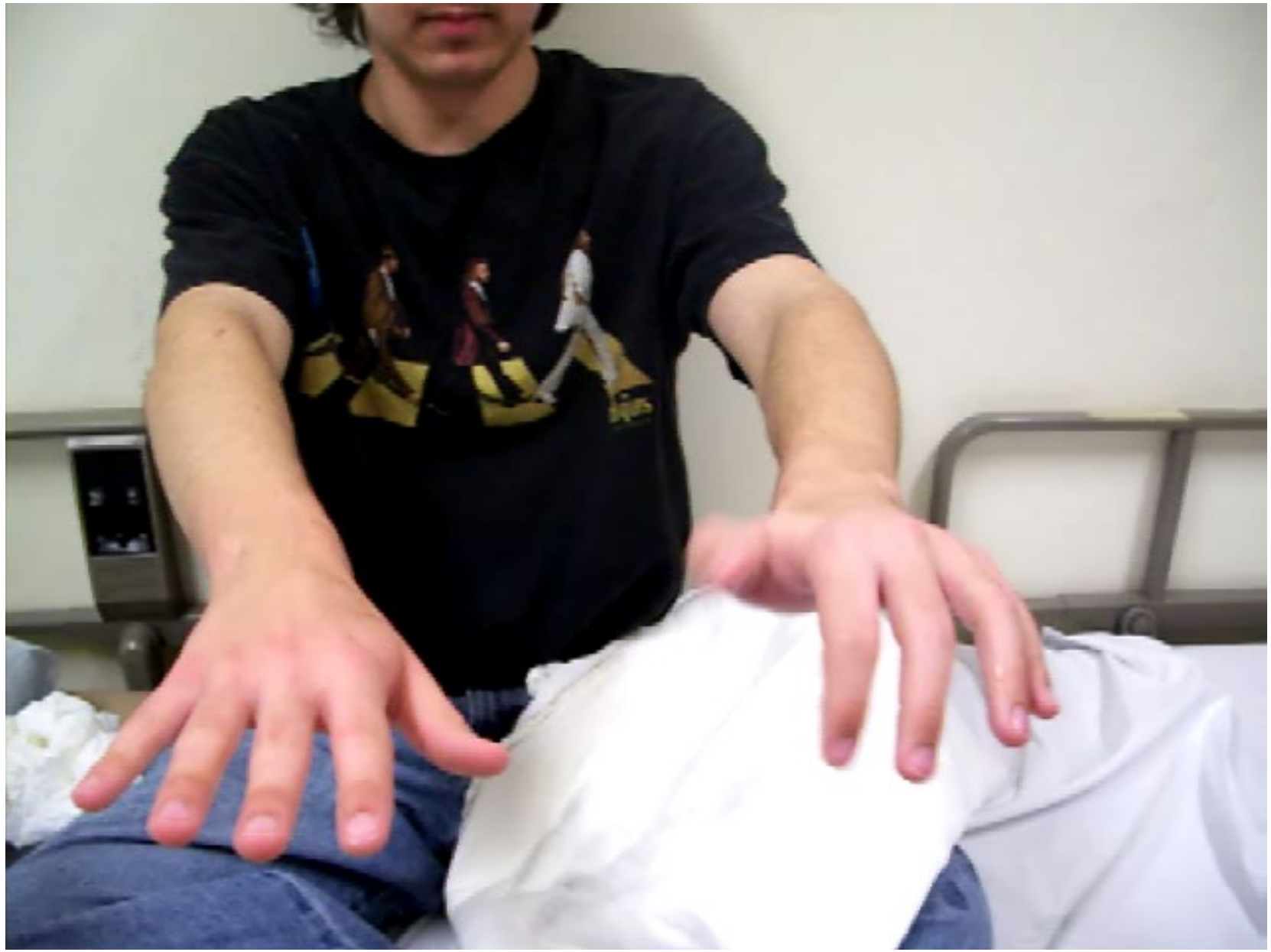
Baseline



Cold Water - Worse



Warm Water - Improved



Diagnosis?

Diagnosis

Hirayama Disease

Differential Diagnosis

- **Hirayama disease**
- Cervical Syringomyelia
- Cervical rib/cervical root compression
- Compressive lesion in the spinal cord
- Upper limb neuropathy/brachial plexopathy
- Juvenile ALS
- MMN (multifocal motor neuropathy)

Key Elements:

- C8-T1 segment involvement
- Symptoms spreading from one side to other
- Worsening with cold and improved by warmth
- No sensory involvement

Hirayama Disease

- Juvenile muscular atrophy of the distal upper extremity
- Juvenile asymmetric segmental spinal atrophy
- Benign focal amyotrophy
- Monomelic amyotrophy

Overview of Hirayama Disease

- Focal anterior horn cell disease affecting C8-T1 and C7 segments
- Unilateral or asymmetric bilateral muscular weakness and wasting of hand and forearm
- Occurs in adolescence, predominantly male
- Progression over initial 1-3 years followed by clinical stabilization
- Described mainly in Asian population, uncommon in the West
- Lack of awareness may lead to initial misdiagnosis

Requirements for the Diagnosis of Hirayama Disease

- Distally dominant muscle weakness and atrophy of forearm and hand
- Onset between ages 10 and early 20s
- Unilateral or unilaterally dominant symptoms and signs
- Insidious onset with gradual progression for the first several years, followed by arrest of progression
- Lack of sensory disturbance
- No symptoms or findings in the lower extremities
- Other diseases (such as syringomyelia, spinal cord tumors, motor neuron disease) must be excluded