Neurological disorders -II

Dr. Abdelrahim A. Sadek

(MD Pediatrics)

MENTAL RETARDATION

Definition

- Limitation in performance that results from sub-average intelligence and adaptive behavior.

Degree of mental retardation

- Mental retardation can be classified according to the intelligence quotient (IQ)

- IQ = $\frac{\text{Mental age X100}}{\text{Chronological age}}$
- Normal IQ= 90-110

Tests For IQ Assessment

- I-Bayley Scales of infant development
- II-Stanford Binet Intelligence Scale
- III-Welchsler Scale

Degree of mental retardation

- Mild MR (85 % of cases): IQ, 50-55 to ~ 70 (Educable)
- Moderate MR: IQ, 35-40 to 50-55 (Trainable)
- Severe MR: IQ, 20-25 to 35-40 (Non trainable)
- Profound MR: IQ, below 20-25

Etiology

- **Prenatal causes**
  - **Genetic**
    - **Chromosomal abnormalities e.g. Down syndrome, Edward syndrome, fragile X syndrome.**
    - **Metabolic diseases e.g. phenylketonuria , galactosaemia and mucopolysaccharidosis**
    - **Congenital brain anomalies e.g. genetic Microcephaly and hydrocephalus.**
    - **Neurodegenerative & Neurocutaneous disorders**

- **Intrauterine**
  - Congenital infections (TORCH).
  - Drugs and toxins e.g. alcohol, phenytoin and cigarettes
  - Radiation
  - Placental insufficiency

Perinatal causes

- During pregnancy: ante partum hemorrhage and toxemia
- During delivery: asphyxia, trauma and intracranial hemorrhage

Postnatal

- Trauma e.g. intracranial hemorrhage
- Infections e.g. encephalitis and meningitis
- Anoxia
- Metabolic e.g. hypoglycemia, Kernicterus
- Endocrinal e.g. hypothyroidism
- Poisoning e.g. lead and copper

Clinical picture

- **Newborn**
  - Abnormal features
  - Feeding and breathing problems
- **Early infancy (2-4 months)**
  - Delayed social development
  - Vision and hearing impairment
- **Late infancy (6-18 months)**
  - Gross motor delay
- **Early childhood**
  - Delayed or difficult speech

- **Preschool children**
  - Delayed or difficult speech
  - Delayed sphincteric control
  - Behavioral difficulties
  - Delayed fine motor skills

- **School age**
  - School failure
  - Underachievement and learning difficulties

Investigations

- **According to clinical possibilities**
- **Radiological:** skull X ray, CT,MRI
- **Laboratory:** Chromosomal, TORCH screen
- **Screening tests for treatable causes**
  - Hypothyroidism: T3,T4 and TSH
  - Galactosaemia: Urine reducing substances
  - Phenylketonuria: urine ferric chloride test

Differential Diagnosis

- Cerebral palsy
- Sensory deficits: blindness and deafness
- Social deprivation
- Some neurological diseases

Treatment

Prevention

- Screening and early treatment of treatable causes
- Vaccination of females against rubella before child bearing period
- Avoid risk factors during pregnancy, labor and after labor
- Avoid further pregnancies in inherited untreatable diseases when risk of recurrence is high.

- Treatment of treatable causes
  - Hypothyroidism: thyroxin replacement
  - Galactosaemia: galactose free diet
  - Phenylketonuria: phenylalanine free diet.
- Supportive treatment for untreatable causes

  -Educational and training programs

  -Treatment of associated problems as epilepsy

  -Emotional support for family

SIEZURE SYNDROMES (CONVULSIONS)

Definition

- Convulsions are one of the most common pediatric emergencies.
- They are a paroxysmal involuntary disturbance of brain function that may be manifested as

  1-Involuntary contraction of muscles

  2-Disturbed conscious level

  3-Sensory or autonomic abnormality

  4-Behavioral abnormalities

Etiology

A-Acute convulsions

1-Febrile convulsions

2-CNS causes

- Infections: meningitis, encephalitis and brain abscess
- Encephalopathy: post vaccination or viral infection, renal or hepatic failure
- Intracranial hemorrhage: trauma, vascular malformation or bleeding disorder
- Brain anoxia

3-Metabolic and electrolyte disturbance

- Hypoglycemia
- Hypocalcemia
- Hypo-or hypernatremia
- Hypomagnesaemia
- Alkalosis
- Pyridoxine deficiency

4-With other diseases

- Gastroenteritis
- Glomerulonephritis

5-Drugs/toxins

- Theophylline, isoniazid
- Snake or scorpion bites
- Acute lead poisoning

6-First fit of epilepsy

B-Chronic recurrent convulsions

- Primary (idiopathic) epilepsy
  - No cause is detected
- Secondary (organic) epilepsy
  - Congenital brain malformations
  - Brain injury: prenatal, natal or post natal
  - Some hereditary metabolic and degenerative diseases

Clinical picture

- look for
  - –Clinical features of the cause if evident
  - –Type of convulsions
    - Tonic: rigid posturing of trunk and extremities
    - Clonic: rhythmic twitching of muscles of face and extremities

- **Tonic-Clonic :is the most common type**
- **Myoclonic: brief jerking (sudden flexion movement) of the body and extremities.**
- **Absence seizures. Attacks of impaired consciousness without falling or involuntary movements.**

Distribution of convulsions

- Generalized: involves all body
- Focal (partial): one extremity or one side of body

Conscious level

- Intact usually in focal convulsions
- Lost usually in generalized convulsions

Duration and frequency

Postictal coma: transient loss of consciousness after prolonged convulsion.

Investigations

Directed according to suspected etiology

- Radiological
  - Skull X ray
  - CT or MRI
- Laboratory
  - Blood electrolytes
  - Renal and liver functions
  - CSF study
  - Blood picture

- Drugs and toxins screen in urine and serum
- Blood gases
- Others according to suspected etiology

- Others
  - ***EEG: is very helpful in diagnosis***.

Differential Diagnosis:

Other conditions mimics convulsions

- Movement disorders
- Vertigo
- Breath holding spells
- others

Treatment

- Treatment of the cause
- Anticonvulsant drugs

Introduction

- Febrile seizures are seizures that occur between the age of 6 and 60 mo with a temperature of 38°C (100.4°F) or higher,
- that are not the result of central nervous system infection or any metabolic imbalance,
- that occur in the absence of a history of prior afebrile seizures.

- A **simple febrile seizure is a primary generalized, usually tonic–clonic,** attack associated with fever, lasting for a maximum of 15 min, and not recurrent within a 24-hr period.
- A **complex febrile seizure is more** prolonged (>15 min), is focal, and/or reoccurs within 24 hr.
- **Febrile status epilepticus is a febrile seizure lasting longer than 30 min.**

- Most patients with simple febrile seizures have a very short postictal state and usually return to their baseline normal behavior and consciousness within minutes of the seizure

- Between 2% and 5% of neurologically healthy infants and children experience at least 1, usually simple, febrile seizure.

- Simple febrile seizures do not have an increased risk of mortality even though they are, understandably, concerning to the parents when they first witness them.

- Complex febrile seizures may have an approximately 2-fold long-term increase in mortality, as compared to the general population, over the subsequent 2 yr, probably secondary to coexisting pathology.

- There are no long-term adverse effects of having 1 or more simple febrile seizures.

- Compared with age-matched controls, patients with febrile seizures do not have any increase in the incidence of
- abnormalities of behavior,
- scholastic performance,
- neurocognitive function,
- attention.
- Children who develop later epilepsy, however, might experience such difficulties.

Febrile seizures recur in

- approximately 30% of those experiencing a first episode,
- in 50% after 2 or more episodes,
- in 50% of infants younger than 1 yr old at febrile seizure onset.
- Several factors affect recurrence risk

Risk Factors for Recurrence of Febrile Seizures

MAJOR

- Age <1 yr
- Duration of fever <24 hr
- Fever 38-39°C (100.4-102.2°F)

MINOR

- Family history of febrile seizures
- Family history of epilepsy
- Complex febrile seizure
- Daycare
- Male gender
- Lower serum sodium at time of presentation

- Having no risk factors carries a recurrence risk of approximately 12%;
- 1 risk factor, 25-50%;
- 2 risk factors, 50-59%;
- 3 or more risk factors, 73-100%.

Risk of Epilepsy

- Although approximately 15% of children with epilepsy have had febrile seizures, only 2-7% of children who experience febrile seizures proceed to develop epilepsy later in life.

- There are several predictors of epilepsy after febrile seizures

Risk Factors for Occurrence of Subsequent Epilepsy After a Febrile Seizure

- Simple febrile seizure 1%
- Recurrent febrile seizures 4%
- Complex febrile seizures (more than 15 min duration or recurrent within 24 hr) 6 %
- Fever <1 hr before febrile seizure 11%
- Family history of epilepsy 18%
- Complex febrile seizures (focal) 29%
- Neurodevelopmental abnormalities 33%

Genetic Factors

- The genetic contribution to the incidence of febrile seizures is manifested by a positive family history for febrile seizures in many patients.
- In some families, the disorder is inherited as an autosomal dominant trait, and multiple single genes that cause the disorder have been identified in such families.
- However, in most cases the disorder appears to be polygenic, and the genes predisposing to it remain to be identified.

- Any type of epilepsy can be preceded by febrile seizures,

- A few epilepsy syndromes typically start with febrile seizures.

Epilepsy syndromes starting by FS

- generalized epilepsy with febrile seizures plus (GEFS+),
- severe myoclonic epilepsy of infancy (also called Dravet syndrome),
- temporal lobe epilepsy secondary to mesial temporal sclerosis.

EVALUATION

History
Exam
Manage acute febrile seizure and acute illness (first aid,midazolam, diazepam, diagnostic tests) as needed.
Determine risk factors for recurrence and estimate risk of recurrence of the febrile seizure

Intermediate or high risk
1. Consider EEG and imaging
2. Consider intermittent oral diazepam or, in exceptional cases that recur, continuous therapy

Low risk
No therapy or investigations are necessary

Counsel parents about risk of recurrence and how to provide first aid and manage fever.

Determine risk factors for later epilepsy

EVALUATION

- Each child who presents with a febrile seizure requires a detailed history and a thorough general and neurologic examination.
- Febrile seizures often occur in the context of

- otitis media,
- roseola
- human herpesvirus (HHV) 6 infection,
- shigella, or similar infections,
- making the evaluation more demanding.

- In patients with febrile status, HHV-6B (more frequently) and HHV-7 infections were found to account for one-third of the cases.

- Several laboratory studies need to be considered in evaluating the patient with febrile seizures

Lumbar Puncture

- Meningitis should be considered in the differential diagnosis, and lumbar puncture should be performed for
- all infants younger than 6 mo of age who present with fever and seizure,
- or if the child is ill appearing
- or at any age if there are clinical signs or symptoms of concern.

Lumbar Puncture

- A lumbar puncture is an option in a child 6-12 mo of age who is deficient in *Haemophilus influenzae type b and Streptococcus pneumoniae* immunizations or for whom immunization status is unknown.

Lumbar Puncture

- A lumbar puncture is an option in children who have been pretreated with antibiotics.
- *In patients presenting with febrile status epilepticus in the absence of a central nervous system infection, a nontraumatic lumbar puncture rarely shows cerebrospinal fluid (CSF) pleocytosis (96% have <3 nucleated cells in the CSF) and the CSF protein and glucose are usually normal.*

CSF RBCs Correction Equation

- WBC in blood x RBC in CSF
- True WBC in CSF =Actual WBC in CSF- ----------
- RBC in blood

- 10000X50
- = 100- ----------=100-0.25=99.75
- 2000000
- 12000x200
- =25000- ----------= 24999.2
  3000000

Electroencephalogram

- If the child is presenting with the first simple febrile seizure and is otherwise neurologically healthy, an EEG need not normally be performed as part of the evaluation.
- An EEG would not predict the future recurrence of febrile seizures or epilepsy even if the result is abnormal.

- Blood studies (serum electrolytes, calcium, phosphorus, magnesium, and complete blood count) are not routinely recommended in the work-up of a child with a first simple febrile seizure
- Blood glucose should be determined in children with prolonged postictal obtundation or with poor oral intake (prolonged fasting).

- Serum electrolyte values may be abnormal in children after a febrile seizure, but this should be suggested by precipitating or predisposing conditions elicited in the history and reflected in abnormalities of the physical examination.
- If clinically indicated (e.g., in a history or physical examination suggesting dehydration), these tests should be performed. A low sodium level is associated with higher risk of recurrence of the febrile seizure within the following 24 hr.

Neuroimaging

- A CT or MRI is not recommended in evaluating the child after a first simple febrile seizure.
- The work-up of children with complex febrile seizures needs to be individualized.
- This can include an EEG and neuroimaging, particularly if the child is neurologically abnormal.

TREATMENT

- In general, antiepileptic therapy, continuous or intermittent, is not recommended for children with 1 or more simple febrile seizures.

- Parents should be counseled about the relative risks of recurrence of febrile seizures and recurrence of epilepsy, educated on how to handle a seizure acutely, and given emotional support.

- If the seizure lasts for longer than 5 min, acute treatment with diazepam, lorazepam, or midazolam is needed for acute management of seizures and status epilepticus).

- Rectal diazepam is often prescribed to be given at the time of reoccurrence of a febrile seizure lasting longer than 5 min
- Alternatively, buccal or intranasal midazolam may be used and is often preferred by parents.
- Intravenous benzodiazepines, phenobarbital, phenytoin, or valproate may be needed in the case of febrile status epilepticus.

- If the parents are very anxious concerning their child's seizures,
- intermittent oral diazepam (0.33 mg/kg every 8 hr during fever)
- or intermittent rectal diazepam (0.5 mg/kg administered as a rectal suppository every 8 hr), can be given during febrile illnesses.

- Intermittent oral nitrazepam, clobazam, and clonazepam (0.1 mg/kg/day) have also been used.
- Such therapies help reduce, but do not eliminate, the risks of recurrence of febrile seizures.
- Other therapies have included continuous phenobarbital (4-5 mg/kg/day in 1 or 2 divided doses), and continuous valproate (20-30 mg/kg/day in 2 or 3 divided doses).

- In the vast majority of cases, it is not justified to use continuous therapy owing to the risk of side effects and lack of demonstrated long-term benefits, even if the recurrence rate of febrile seizures is expected to be decreased by these drugs

- Antipyretics can decrease the discomfort of the child but do not reduce the risk of having a recurrent febrile seizure, probably because the seizure often occurs as the temperature is rising or falling.

- Chronic antiepileptic therapy may be considered for children with a high risk for later epilepsy.

- Currently available data indicate that the possibility of future epilepsy does not change with or without antiepileptic therapy.

- Iron deficiency is associated with an increased risk of febrile seizures, and thus screening for that problem and treating it appears appropriate


Thank you