HISTORY

- 8 Years Old Mutaaraf resident of Taxilla Admitted on 11-10-2008.

PRESENTING COMPLAINTS:
Trauma to multiple body parts since first year of life.
HOPI

- Feeding difficulties
- Frequent burns especially after taking hot liquids
- Frequent trauma to acral and dependent parts, incidental & self inflicted
- Breath holding spells followed by fits requiring antiepileptics
HOPI

- very fond of playing but got repeatedly traumatized
- self injuring behavior
- mutilation of fingers
- scarring of the tongue
BIRTH AND DEVELOPMENT

- Born in Multan, uneventful perinatal history
- 1st year passed uneventfully
- Walking was a bit delayed, rest was normal
- Normal intelligence, was admitted in prep class but left school due to repeated illnesses.
- Hearing, Vision normal
- Speech affected due to tongue loss
FAMILY HISTORY

- INSIGNIFICANT
SOCIO ECONOMIC HISTORY

- Father is Tech II
- Belongs to lower middle class
EXAMINATION
• Height: 115 cm
• Weight: 19 Kg
• B.P: 110/70 mm Hg
• Normal hair, skin and teeth enamel.
• Locomotor system: Difficulty in walking due to self amputation of toes.
  Acro-osteolysis of toes and fingers.
• CNS Examination:
  Sensory system: Intact touch but loss of pain sensations.
  Intact deep pain sensation was observed during the debridement of wounds.
  Rest of the CNS examination was normal.
• Systemic examination was normal
CASE 2

- NOORULAIN
- 3 YEARS
- FEMALE
- RESIDENT OF CHASHMA
- PRESENTED IN OPD ON 24 DECEMBER, 2008
PRESENTING COMPLAINT

- LEFT SIDED TONGUE BITE 1 DAY
- FEVER ON EXPOSURE TO HIGH TEMPERATURE
- H/O ABSENCE OF PAIN SENSATION NOTICED WITH VACCINATION.
- BIRTH HISTORY
- DEVELOPMENT NORMAL
- VACCINATION COMPLETE
- FAMILY HISTORY
EXAMINATION

- WELL ORIENTED, INTELLIGENT CHILD.
- VITALLY STABLE
- 1/3 OF LEFT SIDE OF TONGUE WAS BITTEN WITH BLEEDING.
- SYSTEMIC EXAMINATION UNREMARKABLE
Congenital insensitivity to pain with anhydrosis (CIPA).
Familial dysautonomia (Riley Day syndrome).
Hereditary motor sensory neuropathy.
Lesch-nyhan syndrome.
INVESTIGATIONS

- CBC: Normal
- URIC ACID: within normal limits.
- BIOCHEMISTRY: Normal
- NCS: Normal study
- EMG: Normal study
- Sural nerve biopsy: Refused by the parents of Mutaaraf and was consistent with Congenital insensitivity to pain and anhydrosis (CIPA) in the case of Noor ul Ain.
MANAGEMENT

- SUPPORTIVE MANAGEMENT
- TEETH EXTRACTION
- PADDED SHOES
- COMFORTABLE TEMPERATURE
HEREDITARY SENSORY AUTONOMIC NEUROPATHIES (HSAN):

- HSAN I: Sensory radicular neuropathy.
- HSAN II: Congenital sensory neuropathy.
- HSAN III: Familial dysautonomia or Riley Day syndrome.
- HSAN IV: Congenital insensitivity to pain with anhidrosis.
- HSAN V: Congenital indifference to pain.
CONGENITAL INSENSITIVITY TO PAIN

- Rare condition where a person cannot feel (and has never felt) physical pain
- Cognition and sensation is otherwise normal; for instance they can still feel discriminative touch (though not always temperature and there is no detectable physical abnormality.
- Oral damage (such as having bitten off the tip of their tongue) or fractures to bones.
- Unnoticed infections and corneal damage
• Because the child cannot feel pain, they may not respond to problems, thus being at a higher risk of more severe diseases or otherwise.

• In some people with this disorder, there may be slight mental retardation, as well as an impaired corneal reflex.
CAUSES

• increased production of endorphins in the brain

• In some cases, this disorder can be caused by mutations in the voltage-gated sodium channel SCN9A (NaV1.7).
There are generally two types of non-response exhibited.

*Insensitivity* to pain means that the painful stimulus is not even perceived: a patient cannot describe the intensity or type of pain.

*Indifference* to pain means that the patient can perceive the stimulus, but lacks an appropriate response: they will not flinch or withdraw when exposed to pain.
INCIDENCE

- Due to its congenital nature, the disorder is primarily found in homogeneous societies.
- For example, it is found in Gällivare, a Swedish village in Gällivare Municipality in northern Sweden, where nearly 40 cases have been reported.
A rare genetic disease characterized by absence of reaction to noxious stimuli and anhidrosis.

Point mutations affecting the neurotrophic tyrosine receptor kinase type 1 (NTRK1)/nerve growth factor receptor gene have been detected in CIPA patients.
RILEY–DAY SYNDROME
(Familial dysautonomia)

- Autosomal Recessive
- Incidence-1/10000-1/20000, highest in Eastern European Jews
- Carrier state-1%
PATHOLOGY

- Disease of Peripheral Nervous System
- Reduced Number of
  - small unmyelinated nerve fibers
  - Large myelinated fibers
  - Fungiform Papillae of tongue
  - parasympathetic ganglions in myenteric plexus
- Hypoperfusion of organs and extremities with terminal vessel hyperperfusion
CLINICAL MANIFESTATIONS

INFANTS

- Poor sucking and swallowing, aspirations
- Vomiting crises
- Excessive sweating & blotchy erythema
- Vulnerable to heat stroke
- Episodic Hyperhidrosis
- Breath holding spells & Syncope - common in first 5 years – major motor motor seizures in 40%
OLDER CHILD

- Insensitivity to pain-traumatic injuries
- Corneal & tongue ulcers
- Delayed or clumsy walking
- Reduced tears
- Urinary incontinence
- Arrhythmias
- Delayed puberty
- Short Stature
- Slurred and nasal speech
AUTONOMIC CRIES

- Cyclic vomiting
- Profuse sweating
- Blotchy erythema
- Irritability
- Hypertension
ALLGROVE SYNDROME

- Alacrima
- Achalasia
- Autonomic Dysfunction
- Sensorimotor neuropathy in Adolescence
LABS

- ECG
- CXR
- URINARY VMA
- HVA
- SURAL NERVE BIOPSY-decreased unmyelinated fibers
- EEG
SPECIAL INVESTIGATIONS

- 2.5% methacholine into conjunctival sac
- I/V infusion of Norepinephrine
- Infusion of methacholine
- Intradermal Inj of 1:1000 histamine
- Genetic marker in blood for definitive diagnosis
TREATMENT

- Symptomatic treatment
- Methylcellulose eye drops
- Chlorpromazine and bethanechol for vomiting
- Scoliosis
- Cardiac pacemaker
- Dental management:
  - Night guard.
  - Extraction of teeth.
PROGNOSIS

- Death from hyperpyrexia within first 3 years of life in about 20% of patients.

- Early death due to chronic pulmonary failure or aspiration