Review Article

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Dysmorphism

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Abstract

A *dysmorphic* feature is a difference of body structure. It can be an isolated finding in an otherwise normal individual, or it can be related to a congenital disorder, genetic syndrome, or birth defect. It is estimated that 10% of the pediatric hospital admission involved known genetic conditions, 18% involved congenital defects of unknown etiology and 40% of surgical admissions are of patients with congenital malformations. Dysmorphology is the study of *dysmorphic* features, their origins and proper nomenclature. Dysmorphic features can occur anywhere in the body but are perhaps most often associated with facial features. As a routine part of patient assessment the nurse should screen the major and minor anomalies. The minor anomalies such as clinodactyly or synophrys to severe congenital anomalies, such as heart defects and holoprosencephaly. In some cases, dysmorphic features are part of a larger clinical picture, sometimes known as asequence, syndrome or association. About 2-3% of all children have a majot anomalie. The nurse have to identify the clues to genretic problems by examining the child and considering the physical characteristics of the parents and other family members.

Keywords: Dysmorphism Bell's Palsy; Mocopolysaccharidosis; Global Developmental Delay; Microcephaly and Rickets.

Introduction

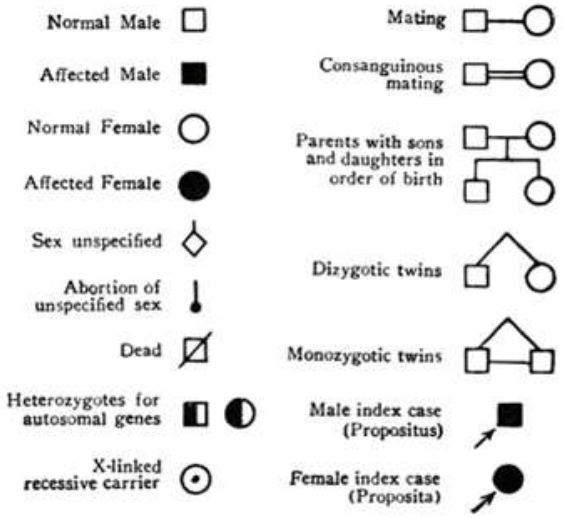
Baby X a 3 years old female child was admitted in the pediatric ward on 9/03/2016 under the pediatric consultant. In the OPD the child was suspected for Bell's palsy with mucopoly saccharidosis and microcephaly. The child was admitted in IPD for further evaluation. Then the child was diagnosed to have dysmorphism under evaluation for Bell's palsy, rickets and mucopoly saccharidosis.

The chief complains was left sided deviation of mouth from 2 to 3 days and Inability to close left eye. Vitals are stable and dysmorphic features are found on head, eyes, extremities, skin, neck, chest and abdomen. The child underwent thyroid function test, X ray of the wrist, CBC, blood chemistry and karyotyping. A stating dose of 10mg of wysolone tablet OD was given for 5 days.

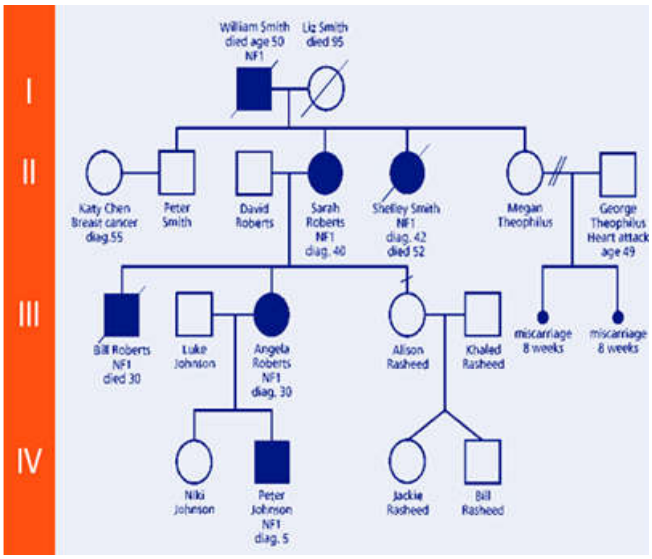
History

There are no congenital abnormalities or chromosomal disorder in the family. No consanguineous marriage between the parents. The child is second in birth order. Her elder sister is 4 years old and seems to be normal. Baby X was full tern normal delivery at home with delayed crying at birth. The ABGAR score was not known. The birth weight was not known. The baby required no hospitalization. Baby X suffering from global developmental disorder at present.

Family Pedigree



Symbols used in Family Pedigree Chart



e.g. Family Pedigree of 4 Generation

Physical Examination

- General appearance: normal body built and Conscious.
- Skin: persistent Mongolian spot.
- Head: Microcephaly positive, prominent occiput, posterior fontanel was still open. Large bulging head, prominent frontal bone. Over riding of sutures.
- Face: coarse facies, left sided deviation of mouth while crying
- Nose: depressed nasal bridge.
- Eyes: hypertelorism. Sclera is pale
- Neck: short neck
- Chest: wide spaced nipple
- Abdomen: abdominal distention present and enlarged. Liver palpable 4 cm
- Extremity: contracture of fingers, short stubby finger. Rocker bottom feet. Wrist widening . over riding of toes.
- Vital signs: Temp: a febrile, Pulse: 70beats/ minute, Repiration: 20 breaths/ minutes and BP: 120/80mm/hg
- ***Anthropementry:***
  - Height: 72 cm
  - Weight: 8.2 kg.
  - Head circumference: 42.5 cm
  - Mid arm circumference: 16cm
  - Chest circumference: 50cm
  - Abdominal circumference: 52cm

Degree of Malnutrition

58% third degree malnutrition according to Gomez classification.

Growth and Development

Growth and development	
Book picture	**Patient picture**
Physical or Biological Development	
• Weight : Approximately: 14 kg • Height :Approximately 85 to 95cm • Pulse: 95 beats/mt • Respiration :25 + breaths /mts • Blood Pressure :100/67 + 24/25	• 8.2 kg • 72cm • 70b/m • 20 breaths/m • 120/80mm/hg
Gross Motor	
• Walks a straight line. • Walks on tiptoes • Runs without looking at feet	• She was able to sit with support at 1 years.

- Catches ball with extended arms.
- Kicks a ball
- Jumps from a height of several inches.
- Ride tricycle using pedals turns wide corners.

Fine Motor

- Builds a tower of 9-10 blocks
- Copies a circle
- Shows preference for handedness
- Can help with simple house hold task.

Self Care

- Can put on coat without assistance.
- Can Undress self most instances
- Toileting and grooming skills.
- Can pull pants up and down.
- Can go to toilet alone
- Brushes teeth with help.

Psychosocial development :

- Beginning development of sense of initiative.
- Ego Centric
- Known own sex

Psycho Sexual Development :

- Phallic Stage (3 – 6 Years)

Spiritual development :

- Intuitive – Projective faith.

Cognitive development

Sub stage - I :

- Pre-conceptual (2 – 4 Years) characterized by language acquisition

Moral Development

- Pre Conventional Morality Stage 1 (2 - 3 Years)

Receptive Language Development

- Can obey two prepositional commands. (ie – On, Under)

- Roll over at one year.
- Not able to stand without support

- Able to hold the object in both hand

- Completely Lacking

- Not seen initiation

- Still oral stage
- Undifferentiated

- Sensory stage
- Egocentric
- Can say bisyllable word (mama)

Disease Condition

Topic	Book Picture	Patient Picture
Definition	A **dysmorphism** is an anatomical malformation *have facial dysmorphism and other s abnormalities. It is a* difference of body structure. It can be an isolated finding in an otherwi individual, or it can be related to a congenital disorder, genetic syndrome, or birth defect.	
Incidence:	➢ Major congenital anomaly • At birth : 2-3% • At 5 years 4-6% ➢ Minor congenital anomaly: • At birth 15 %	• Seen from newborn
Classification	• Deformation (unusual mechanical pressure during developmental period especially during last trimester) • Disruption (abnormal cellular organization) • Dysplasia (affect structure that has been undergone normal development and growth in utero) • Malformation (intrinsic abnormalities of development in body structure during prenatal period)	• Disruption

Causes

- Congenital disorder
- Genetic syndrome
- Birth defects
- Teratogen exposure
- Environmental
- Twining
- Unknown
- Multifactorial
- Rickets, Bell's palsy
- Mucopolysaccharidosis
- Hypoxic ischemic encephalopathy

Types of anomalies

- Minor (requires social and medical implications, rarely requires surgical implication)
- Major (mostly cosmetic significance)
-

Pathophysiology

- Pathophysiology is not well understood

Due to congenital, genetic and birth defects

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Development of dysmorphic features

E.g for cranisynostosis:

Autosomal dominant gene → mesanchymal blasmtoma

Hyperthyrosidm → acelerated ossicious maturation → craniosynostosis

Microcephaly → lack of growth stretch across sutures

Clinical feature

Head:

- Asymmetric head
- Fontanel too large or small
- Microcephaly
- Prominent or flat occiput
- Craniosynostosis
- Micrognathia (small jaw)
- Post fontanel still open
- Microcephaly
- Prominent occiput
- Craniosynostosis

Ears:

- Ear tags
- Posteriorly rotated
- Hearing loss
- Low set or malformed ears
- Ear tags

Eyes:

- Blue sclera
- Up or Down slanting eyes
- Extreme hyperopia or myopia
- Hypertelorism or hypotelorism
- Hypertelorism (widely spaced eyes)

Skin

- Extremely loos or thin skin
- Hirsutism
- Hyperelastic skin
- Leaf shaped white markings
- Webbing between finger and toes
- Persistent Mongolian spot
- Webbing between finger and toes

Mouth:

- Cleft lip with or without cleft palate
- Large or small tongue

- Early loss of teeth
- Late eruption of teeth
- Thin upper lip
- Mouth is deviated to left side
- Late eruption of teeth

Extremities:

- Contracture of the hand feet, toes
- Arachnodactyly (long finger or toes)
- Bradytactyly (small finger or toes)
- Camptotactyle (permenant flexion of finger or toes)
- Clinotactyle (curved fingers or toes)
- Extremely long or short extremities
- Edema on the extrimities
- Hypospastic
- Hypotonic
- Loose joints
- Polydactyle or syndactyle
- Single palmar cleases
- Rocker bottom feet
- contracture of fingers,
- Short stubby finger.
- Rocker bottom feet.
- Wrist widening.
- Over riding of toes.

Others:

Investigation

- Family history
- Physical examination
- Blood investigation
- Karyotyping
- TFT
- X ray
- USG
- Blood chemistry (Metabolic studies)
- Hb: 9.3gm/dl
- Blood chemistry: Na: 134.3 mmol/l, K+: 4.02 mmol/l, Cl- : 102 mmol/l, and Ca++: 1.03 mmol/l
- Usg abdomen to rule out KUB anomalies.
- X ray of the left wrist with tipoff the finger.
- Thyroid function test.
- Karyotyping to rule out genetic abnormalities.
- Complete blood count.

Treatment

- Constructing the pedigree and analyzing of the padigree
- Review patient past records and prenatal history
- clinical assessment:
- Visual assessment
- Measurement
- Extended family
- Genetic counseling\
- Follow up
- Clinical assessment was done
- Genetic screening was done
- Genetic counseling provided
- Started with tablet wysolone 10 mg OD for 5 days.

Nursing Management

A. Assessment

The assessment consists of a comprehensive evaluation of the shortcomings and strengths associated with the adaptive skills; communication, self-care, social interaction, use of facilities in the community self-direction, health care and safety, functional academic, recreational skill formation, and tranquility.

B. Nursing Diagnosis

1. Impaired growth and development related to cognitive dysfunction.
2. Impaired verbal communication related to cognitive dysfunction.
3. Impaired Social Interaction related to difficulty speaking / social adaptation difficulties.
4. Interrupted family processes related to having a child with mental retardation.
5. Self care deficit related to changes in physical mobility / lack of developmental maturity.
6. Risk for injury related to aggressive behavior imbalance of physical mobility.

C. Intervention

1. Assess the factors causing impaired child development.
2. Identification and use of educational resources to facilitate optimal child development.
3. Genetic referrals and counseling- After gathering assessment data that incorporate the genetic concepts, the paediatric nurse is able to partner with children and their families by initiating a referral to genetic specialist if there are indicators for a genetic referral
4. Psychological issues :Provide psychological support and guidance throughout the care of the child..
5. Provide consistent care.
6. Increase communication verbal and tactile stimulation.
7. Give simple instructions and repeat.
8. Give positive reinforcement on child outcomes.
9. Encourage children to do their own maintenance.
10. Difficult child behavior management.
11. Encourage children to socialize with the group
12. Create a safe environment.

D. Education on Parents

1. Each stage of child development for ages.
2. Support parental involvement in child care.
3. Anticipatory guidance and management face a difficult child behavior.
4. Inform existing educational facilities and groups.

E. Expected outcome

1. Children to function optimally the relevant level.
2. Families and children are able to use coping with challenges due to disability.
3. Families are able to obtain the resources community facilities.

Prognosis	Depends up on the causes and type of dysmorphic features.	Baby is active but dysmorphic features are not shown any favorable prognosis

Discussion

Dysmorphsim if a Greek word means badness of form. This refers to the malformation and abnormality in the shape and size of the body. It can be facial or structural. Baby X nomenclature of the present health problems is developmental and genetic in nature with multiple major anomalies along with that deformity Syndrome is seen. The baby is given wioth stating dose of tablet wysolone 10mg/day for 5 days and was discharged with genetic counseling to the parents and asked to come for follow-up.

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