

Cornelia De Langue Syndrome

Child 14 months old, Master Sunder, was brought to me for delayed development.

The main complaints were of poor weight gain [5.5 kg]. Slow Mental development and slow milestones- no dentition or speech till date..

When I saw the patient for the first time, his face, expressions and movements were like that of a small baby monkey. He had plenty of hair all over his body +10. He had bushy eyebrows, long eyelashes, a small nose and thin drawn up legs - features evident of Hirsutism ++.

LIFE SITUATION:

Mother had tremendous tension and worry during pregnancy. Main cause-Future Dowry. The mother came from a community where a girl has to be given heavy dowry; the mother therefore wanted a male child. She always worried what if she got a girl. Her sleep was disturbed 3+ and she had severe complaints of acidity 3+ and vomiting 3+ during this period. She would start in sleep, worry 3 at night and often dreamt that someone was harming her. She felt neglected during pregnancy as there was not much of co-operation from her husband.

(In the past, she had got an amniocentesis done and had abortion since it was a female child)

PERSONAL HISTORY

Appetite-Less

Craving-Sweets +3

Bowel-Constipated, passes stool every alternate day.

Feels better after burping; otherwise vomits.

Sweat-+++ fan is must, likes air conditioning

Urine-Nil

Sleep-Gets up with a startle; when patted goes back to sleep.

Thermal modality:Hot

Bath-Tepid. Cover-Aversion

Weather-Comfortable in winter

PAST HISTORY

Birth weight 2.2 Kg.

P/H/O. Bronchitis, cold, cough, constipation frequently

Cleft palate-operated 3 months back.

Tongue tie-operated

F/H: Mother-Cold++. MGF-Diabetes

CLINICAL EXAMINATION

O/E Flat head, fontanelles open.

Hair +10- face, back and all over body, Eyebrows bushy. Hirsutism.

Undescended testes ++ bilateral.

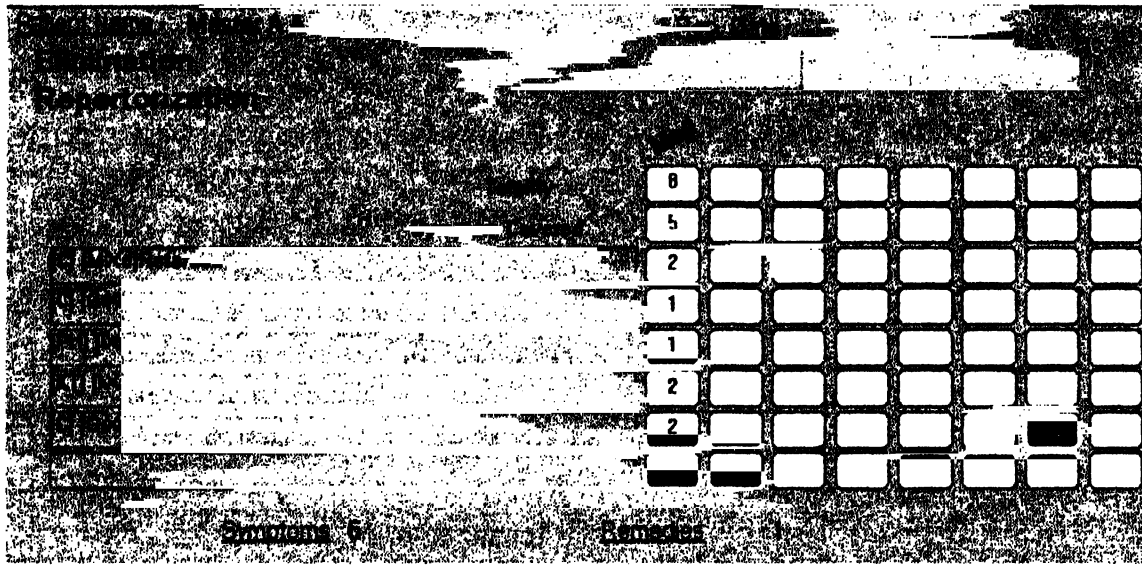
DIAGNOSIS of this syndrome in the case of Sunder was based on following criteria:

1. Low birth weight.
2. Facial features-like that of monkey, small head, hair growth, eye brows bushy etc.
3. Hirsutism.
4. Undescended testicles.
5. Congenital anomalies-cleft lip and palate
6. Language delay.
7. Growth retardation, no teething, etc.
8. Behavioural problems.
9. Poor sleep and appetite.
10. Mental retardation.

DIAGNOSIS:

Congenital Anomalies, Microcephaly. Cornelia De Lange Syndrome.

DR. JAWAHAR SHAH
8, New Hari Niwas, Dattatray Road
Santacruz (W), Mumbai-400054
Tel: 022-2661 4532, 2661 6561
Fax: 022-2660 0204
Email: hompath@vsnl.com
Web: www.hompath.com



1/6/93 *Medo 30 3P N* SL pill TDS
 7/6/93 Playful Active +
 Follows object and movements
 Makes and attempt to get up. Rx SL
 14/6/93 Recognizes small objects like mango, etc.
 Activity>> Rx SL
 21/6/93 Tooth eruption ++ Rx SL
 30/6/93 Turns, recognizes +5 Active on rolls++
 Wants to go out. Smiles
 When taken out and brought back,
 sees home and start crying
 Recognizes and shows finger to go out
 Extra hair has started falling
 Rx SL 1-1 for 3 months
 11/11/93 Improvement ++++
 Can recognize people / objects,
 mixes around with everyone.
 Can do many things on his own. Rx SL
 12/12/94 Improvement continues. Hair fall +5
 Can recognize more objects
 Differentiates taste. Rx SL
 21/4/2001 Patient is alive till last known with consider-
 able improvement in over all performance mentally and
 physically.
 Patient is 9 yrs old, weight of child is 12 kg and mental
 age is of four and a half years.

Following improvement observed in the child:

- Can understand instructions in three languages English, Gujarati and Hindi.
- Understands commands well.
- Recognizes voices of family, friends and makes an attempt to speak on phone.
- Can hold pencil and scribble.
- Makes signals and gestures for whatever he wants
- Likes to keep things in order.
- Sensitive and moody in nature.
- Loves to mix with older people.
- Has developed a good coordination between picture and words.
- Sleeps well.
- Loves to listen to music.
- Can play with beats and rhythms.
- Can play with others.
- Desire to travel, to go out, meet people.
- Shows way out of the house.
- Recognizes good clothes and appreciates with positive gestures.
- Loves cold drinks, Mac Donald's food and insists mother to give such food.
- Whenever there is a pit, understands and moves accordingly.

