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The year 2010 was a landmark year for IAP chapter on Growth, Development and Behavioral Pediatrics. On 18 June 2010 at the Institute of Child Health, Kolkata the four modules on Growth Monitoring, Developmental Screening, Autism and Behavioral Problems were given the final shape. It started in 2008 when the chapter took the initiative to develop modules on four important issues related to growth, development and behavioral pediatrics. Subsequently a task force was formed and two sessions were held in 2008 and 2009 as the preliminary steps to develop the modules. Finally the task force came together and gave final shape to the modules. The main purpose behind development of these modules was to orient and train the members and post graduate students in important issues related to growth monitoring, developmental screening, autism and common behavioral problems. Dr Dilip Mukherjee, Dr K N Agarwal, Dr Ksh Chourjit Singh, Dr M K C Nair, Dr Nandini Mundkur and Dr Sukanta Chatterjee are the advisors of these modules. The members are Dr Anjana Thadhani, Dr Anju Seth, Dr Jaydeb Ray, Dr Jaydeep Choudhury, Dr Madhuri Kulkarni, Dr Monideepa Banerjee, Dr Ranjana Chatterjee, Dr S N Parida, Dr Sabina Ahmed, Dr Suchit Tamboli, Dr Swapna Mittal and Dr Zafar Meenai. President of Central IAP 2010, Dr Deepak Ugra, President Central IAP 2011, Dr T U Sukumaran and Secretary General, Dr Tanmay Amladi are the Ex-officio members.

The module on Growth Monitoring focuses on growth assessment, anthropometric measures of weight, length, height and head circumference, pitfalls in measurement and assessment of puberty. It also highlights on interpretation of anthropometric data, growth chart, concept of centiles, Z score, SD, BMI, types and plotting on charts, interpretation, prediction of adult height based upon current growth calculation of genetic potential and comparing whether a child’s growth is commensurate with it understanding the significance of bone age, differentiate between various normal and pathological growth patterns.
The module on Developmental Screening demonstrates on identifying the neonates, infants and children at risk for developmental delay, recognize the flag signs of development delay, understand the etiological factors and co-morbidities in developmental delay, assess the developmental delay in various domains, plan management of developmental delay and understand the preventive aspects of developmental delay.

The module on Autism stresses on identifying children at risk for autism, identify the red flags in autism and speech delay, recognize the deviant features in development in autism, diagnose a child with autism, make appropriate referrals, emphasize the need for early detection and early intervention.

The module on Behavioral Problems deals with childhood developmental disorders including pervasive developmental disorders, attention-deficit and disruptive behavior disorders, eating disorders, habit disorders, somatoform disorders, psychotic disorders, substance misuse, emotional disorders like phobias, school phobia, OCD, anxiety states, separation anxiety, panic disorders and selective mutism and conduct disorders like stealing, defiance, aggression and anti-social behavior.

The plan is to conduct small workshops on these modules at various places. The sincere efforts of the committee will be successful only if the message is passed on many members and then utilized in day to day practice.

Jaydeep Choudhury  
Editor-in-Chief
Issues in Linear Growth in Children

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Growth is the sole indicator of health. Growth is influenced by nutrition, illness and environment. Approximately 10% of children may present with growth problems.

Growth disorders may be due to excessive shortness (short stature), excessive tallness (Tall stature), fatness (obesity), thinness and problems of puberty. Thus growth disorders may be classified according to the following.

1. Stature – Short or tall
2. Weight – Obesity or thinness
3. Puberty – Delayed, early or secondary characters.

**Short Stature**

A short stature is defined as

(i) Height less than 2 Standard Deviations (2SD), or

(ii) Height is less than 3rd percentile of standard for child’s age and MPH (Mid-parental height) or

(iii) Growth velocity is low (< 5-6 cm/year from age of 3 years till onset of puberty) based height measurements (if measurements likely to be accurate, and are over a period of at least 6-12 months).

Diagnosis of the child who is short and growing slowly comprises the whole of pediatrics and is not just confined to endocrinal disorder. So it is a very vast field. The parents would be worried about the child’s height when,

(i) The child is too short for his parents (ie, below 2SD).

(ii) Growth prediction is too low (by mid-parental height [boys= (father + mother +12)/2; girls= (father -12+ mother)/2]), the predicted height = mid-parental height ± 8 cm. The MPH should be plotted on the proper percentile and this will be the centile the child is likely to follow.

(iii) History does not account for the present situation.

(iv) Abnormal body proportion (abnormal upper/lower segment ratio or span/height ratio).

(v) Deceleration of the growth velocity.

Short stature is classified as shown in figure1.

**Normal Variations of Growth**

**Familial genetic short stature:**

It is the most common cause of short stature is due to low genetic endowment. Here the parents and/or close relatives are short. Child’s projected adult height falls within 2 SD of the MPH.
Here bone age is same or within 2 years of the chronological age. The growth velocity is normal, physical examination and sexual maturation is normal. These children require no treatment. The parents and the child should be reassured. But pathological short stature in parents should be ruled out.

Bone age (BA) should be done mainly to identify overlapping constitutional delay.

Bone age = Chronological age (CA) > Height age (HA). Final Height is in consonant with the parental height and likely to be shorter than the peers.

**Constitutional delay in growth (CDGP):**

They are also called late matures. It is more common in boys. Male: Female = 10 : 1. Puberty and pubertal growth spurt is delayed by > 2 yrs. Periods of slow growth occurs in the first 3 to 4 years of life, so the height falls below the 3rd centile, then the growth velocity is normal for age, growth pattern runs parallel to the normal growth but below the 3rd centile. Height and bone age are usually delayed by 2 to 4 years.

There is history of delayed growth and sexual development in parents and other family members. Final adult height is reached around 20 years, and is within low normal range. Sexual development and fertility is normal. A type of transient, functional hypopituitarism ("lazy pituitary") gets corrected as puberty progresses.

Growth hormone levels, if measured, are normal. BA = HA < CA.

Parents and the child must be explained...
that the child would continue to grow for a longer period than usual and ultimately will reach normal adult height. Testosterone can be used in older boys when delay in puberty becomes psychologically unbearable.

**Pathological Short Stature**

Proportionate and heavy:
Generally endocrinial etiology is present.
(i) Congenital GH deficiency (?midline defects, perinatal asphyxia).
(ii) Acquired GH def. (tumors, trauma, post-infectious).
(iii) Hypothyroidism.
(iv) Cushing syndrome
Investigations according to the cause. TSH, FT4, IGF-1 and IGFBP3, bone age determination. For Cushing’s syndrome consider overnight dexamethasone suppression test or 24 hour urinary cortisol.

Proportionate and thin:
The list is endless.
(i) GI loss – Malabsorption, IBD, celiac disease.
(ii) Renal – RTA, nephrogenic DI, chronic renal failure.
(iii) Cardiovascular – Shunts, failure.
(iv) Endocrinal – Diabetes mellitus, diabetes insipidus.
(v) Pulmonary – Cystic fibrosis, asthma.

Disproportionate short stature:
An abnormal upper/lower segment ratio or span/height ratio indicates disproportionate short stature. Skeletal dysplasia is one of the important investigations in this group.

*Short trunk* – In short-trunk dwarfism (Mucopolysaccaridosis etc), span is greater than height, and US/LS ratio is decreased

*Short limb* – In short-limbed dwarfism (Achondroplasia etc), the height is greater than arm-span, and US:LS ratio is high.

**Dysmorphic:**
These are generally syndromic. Turner syndrome, Down syndrome, etc. Karotyping and geneticist help is needed.

During the assessment of a short child the following thing has to be taken
(i) Full medical and social history
(ii) Accurate measurement of height of child and parents (for accurate height, growth velocity and mid parental height).
(iii) Good observation and thorough clinical examination and investigation
(iv) Bone age, chronological age, height age and body proportion assessments((upper / lower segment ratio and arm-span / height ratio)
(v) Karyotyping in specific cases
(vi) Specific investigation according to the systemic disease.
Biochemical investigation and Endocrinal tests accordingly.

**Outcome and treatment:**
The outcome and treatment of the short stature will depend upon the cause.

(i) Familial genetic short stature, constitutional delay in growth (CDGP) and most causes of intrauterine growth retardation – No specific treatment, only reassurance.

(ii) Skeletal dysplasias – Limb lengthening with limited benefit.

(iii) Specific therapy for chronic systemic disorder, emotional deprivation is associated good catch up.

(iv) Growth hormone is indicated in children with growth hormone deficiency.

(v) Hypothyroidism should be managed with replacement by thyroxin.

**Prognosis:**

(i) Normal and short – Do not need treatment.

(ii) Short due to intrinsic causes – Cannot be treated.


**Tall Stature**
Tall stature and excessive overgrowth syndromes represent physical development in excess of 2 standard deviations (SD) above the mean for the person's age and gender. The incidence varies greatly depending on the etiologies. Following are the causes of tall stature.

**Normal variation:**

(i) Familial tall stature

(ii) Increased caloric intake

**Pathologic:**

(i) Marfan syndrome

(ii) Homocystinuria

(iii) Klinefelter syndrome

(iv) Precocious puberty

(v) Cerebral gigantism (Soto's syndrome)

(vi) Pituitary gigantism and acromegaly

**Approach to tall stature without abnormal features**
If growth velocity is normal, with advanced bone age it may be due to familial tall stature, or due to increased caloric intake. Normal look, increased growth velocity and without sign of pubertal growth could have growth hormone excess or hyperthyroidism. Approach to tall stature without abnormal features is shown in figure 2.

**Familial tall stature:**
Familial tall stature is the commonest cause of excessive height. Height of these individuals is usually above ninety-fifth percentile since early childhood, and their height velocity is within normal limits. Physical
TALL STATURE

without abnormal feature

Growth velocity

Normal

Familial tall stature
Increased caloric intake (obesity)

Increased caloric intake or hyperthyroidism.

Investigate

Increased

Is there any sign of puberty?

NO

YES

Fig 2. Approach to tall stature without abnormal features

examination and bone age are normal. One or both parents are usually tall. Diagnosis can be made from history including family history, normal physical examination and bone age, and growth record.

*Increased caloric intake:* Many children who are overweight because of increased caloric intake also have height that is more than ninety-fifth percentile for age and gender. In this circumstance, increased caloric intake contributes to increased height.

*Without abnormal feature with increased growth velocity:* 

*Precocious puberty* – In children with
precocious sexual development, excessive androgen or estrogen secretion causes accelerated growth and evidence of either virilization or feminization. Bone age is advanced. Although accelerated growth results in tall stature in childhood, premature closure of epiphyses limits growth period, producing attenuation of final adult height.

Approach to tall stature with abnormal features is shown in figure 3.

**Approach to tall stature with abnormal features not disproportionate**

**Cerebral gigantism (Soto’s Syndrome):**

Autosomal-dominant disorder, birth length is typically above ninetieth percentile. Characteristic findings include macrocephaly, prominent forehead and jaw, hypertelorism, down slanting palpebral fissures, high-arched palate, developmental delay, seizures, and mental retardation. Although growth may be accelerated in first years of life, final height may not be excessive. Growth hormone secretion and other tests of endocrine function are normal. Physical findings and growth pattern confirm diagnosis.

**Pituitary gigantism and Acromegaly:**

Pituitary tumors (usually eosinophilic or chromophobe adenomas) and eosinophilic hyperplasia without adenoma produce excessive amounts of growth hormone, which cause gigantism. Final height is 213–275 cm (83–108 in.). Acral changes include coarse facial features with enlargement of nose, ears, and jaw; soft tissue swelling; and striking increases in size of hands and feet. Screening test for growth hormone excess is increased serum concentration of insulin-like growth factor-1. CT or MRI defines location and extent of lesion. Histologic diagnosis is definitive.

**Approach to tall stature with abnormal features disproportionate**

**Marfan syndrome:**

Autosomal-dominant disorder caused by mutations in fibrillin-1 gene located on chromosome15. Most important clinical features are in skeletal, cardiovascular, and ocular systems. Skeletal features include increased height, disproportionately long limbs and digits, joint laxity, scoliosis, thoracic lordosis, pectus excavatum or carinatum, and narrow high-arched palate. Cardiovascular findings include mitral valve prolapse, mitral regurgitation, aortic root dilatation, and aortic regurgitation. Subluxation of lens (usually upward) and myopia are characteristic ocular findings. Clinical findings and positive family history when available confirm diagnosis.

**Homocystinuria:**

Most common form of this autosomal-recessive disorder is due to decreased activity of enzyme cystathionine beta-synthase, which catalyzes formation of
cystathionine from homocysteine and serine. Gene locus has been mapped to chromosome 21q22.3. Clinical features include long limbs, narrow hands with long fingers, limited joint mobility, subluxation of lens (usually downward), osteoporosis, thromboses of arteries and veins, and developmental delay. Some children with this disorder are taller than average. Increased serum concentrations of homocysteine and methionine are diagnostic. Enzyme assay of cultured fibroblasts is confirmatory.

**Klinefelter syndrome:**
Underlying defect in Klinefelter syndrome is presence of extra X chromosome in a boy. Diagnosis is usually made after the time of expected puberty. These individuals have small testes, decreased facial and axillary hair, and often gynecomastia. Karyotype 47, XXY is diagnostic.

**Diagnostic approach to tall stature**

**History and examination:**

1. Family history
2. Parents' heights and puberty ages to judge child's growth relative to genetic potential (midparental target height); family history of chronic disease or endocrinopathy.

**Fig 3.** Approach to tall stature with abnormal features.
3. History of pubertal signs.
5. Symptoms of impaired secretion of gonadotropins and thyrotropin corticotropin, or hyperprolactinemia.
6. Vital signs,
7. Anthropometry (height, weight, sitting height)
8. Features of acromegaly – Coarse face, broad nose, separate teeth, increased mandible growth, dorsal kyphosis, enlarging hands and feet with thickening fingers and toes, visual field defects

Laboratory tests:
1. Bone age X-ray to predict final height.
2. IGF-I is most sensitive screen for GH excess.
3. Karyotype in male patients.
4. Thyroid function tests.
5. Gonadotropins and sex hormone levels if puberty abnormal.
6. Glucose suppression test – Suppress serum GH to <5 ng/dL after 1.75 g/kg oral glucose challenge, gold standard for verifying GH excess.
7. MRI evaluation of pituitary mandatory if evidence of GH excess.

References:
More and more smaller and sicker neonates are surviving over the past three decades with improved treatment modalities. Concurrent advances in reproductive technology have resulted in increasing numbers of preterm and multiple births. These high risk neonates account for infant mortality and morbidity and evidence shows that despite the neonatal interventions very low birth weight (VLBW) remain at a substantial risk for long-term morbidity including cerebral palsy, mental retardation, developmental delay, school problems, behavioral issues, growth failure and overall poor health status\(^1\).

Outcome refers not only to initial 2 years but also includes school age, adolescence and adult life. Focus is moving from very VLBW to extremely low birth (EBLW) and now to extremely immature infants (< 25 weeks gestation) because with decreasing birth weight and gestational age more severe and different morbidities have become apparent\(^2\).

Last two decades have seen tremendous interest in neonatal intensive care all over India but sadly very little attention has been paid to less glamorous care such as systematic follow up of high risk infants. Some of the most elegantly done follow up studies in India are ‘Pune Children’s Studies’ by Chaudhari et al on children born in late eighties\(^3\). However neonatology in India has taken great strides in last 10 years and hence there is a need to continue to correlate neonatal risk factors and interventions with long term outcomes to help us improve our strategies and also understand cost-efficacy of this expensive care. This article is an overview of predictors of long term outcome of a high risk neonate who is exposed to perinatal stress.

**Antenatal Predictors**

*Ultrasound and Doppler examination:*

Abdominal circumference below fifth percentile predicts immediate outcome. The best antenatal predictor of poor outcome was abnormal venous flow, based on the DV Doppler. At five years of age 54% of children born after raised umbilical / cerebral or U/C ratio were functioning below the expected level, compared to 20 % of children born with normal U / C ratio. There is association between abnormalities in the fetal aorta Doppler studies and minor neurological disability\(^4\).

**Maternal diseases and antenatal**
**Steroids:**
Preterm born to mother with preeclampsia do better because they have less IVH and PVL but in contrast preterm infant exposed to chronic choriamnionitis and elevated pro-inflammatory cytokines in amniotic fluid have increased risk of PVL and Cerebral Palsy. Single dose of antenatal steroids is well known to prevent RDS however fetal exposure to multiple/weekly courses of steroids is known to result into smaller head size. Multiple prenatal factors like infection, metabolic, anoxia, toxic, genetic, infarction etc may result into spastic Cerebral Palsy.

**Natal Predictors**

**Birth asphyxia:**
Drage et al. found that 5 minutes Apgar score gave a better prediction for neurological abnormality later on and post asphyxial encephalopathy grade III could predict significant morbidity and sequelae among survivors. An interesting finding from a large prospective NCPP study at USA and case control study done at western Australia have conclusively shown that not more than 10% of cerebral palsy cases were at best related to birth asphyxia. Sigmund Freud who suggested that the baby may have asphyxia primarily because of a prenatal structural or metabolic insult to the baby’s brain and that asphyxia is only a middle episode of a long drama starting with intrauterine asphyxia and presenting with a low Apgar score and neurological deficit later on.

**Postnatal Predictors**

**Birth weight:**
Birth weight > 800 grams seems to be one of the good predictors of intact survival. However the developmental outcome of ELBW infant is determined by a complex interaction of medical and environmental factors acting on the developmentally vulnerable premature brain. 7-17% has neuro sensory impairment and 13-37% delays in cognitive function and also quite high rate of behavioral problems. Since 2000 there is net increase in survivors without impairment however the rates of subnormal cognitive function continue to remain unchanged. VLBW and ELBW children have more respiratory illnesses leading to hospital readmissions and other health problems in initial years but those extremely premature entering adulthood showed no significant differences between the ELBW adults and term-born controls with regards to rates of high school graduation, college enrollment permanent employment and independent living status.

**Prematurity:**
The greater the immaturity and the lower the birth weight, the greater the likelihood of intellectual and neurologic deficit. 50% of 500-750g infants have a significant neurodevelopmental impairment (blindness, deafness, mental retardation, cerebral palsy.)
With increasing gestational age, survival rates increase to approximately 15% at 23 week, 56% at 24 week, and 79% at 25%. The survival of infants <24week gestation, weighing <750g with a 1- min Apgar score <3 is 30%. These infants are also at risk for poor neurodevelopmental outcome. Birth-weight – specific neonatal diseases such as grade III IVH, severe group B streptococcal pneumonia, and pulmonary hypoplasia also contribute to a poor outcome.

**Cerebral palsy:**

Children with ‘transient dystonia’ had an excess of school problems at a mean age of 6.7 years.

The developmental outcome of ELBW infant is determined by a complex interaction of medical and environmental factors acting on the developmentally vulnerable premature brain. 7-17 % has neuro sensory impairment and 13-37 % delays in cognitive function. Disability rate as high as 49 % is reported. Spastic diplegia is most commonly associated with pre-term birth.

Cerebral Palsy is caused by not only prematurity or low birth weight but a series of life threatening medical events. Spastic Cerebral Palsy is caused by prenatal factors like infection, metabolic, anoxia, toxic, genetic, infarction. Ataxic cerebral palsy is caused by perinatal cause such as anoxia. Atonic cerebral palsy is caused by post natal factors such as toxin, trauma, infection.

Many surviving LBW infants have hypotonia before 8 mo corrected age, which improves by the time they are 8 mo-1yr old. This transient hypotonia is not a poor prognostic sign.

School age outcome of premature children consists of significantly poorer intelligence, achievement and higher rates of behavioral problem. These scores are directly proportional to the degree of immaturity. Problems can be poor vocabulary skill, significant delay in reading, spelling and mathematics. The problems that appear to be related to academic failures include deficits in attention; memory and behavior are affected in preterm, ELBW babies.

**Hypoxic ischemic encephalopathy:**

Ataxic cerebral palsy is caused by perinatal cause such as anoxia. It causes severe motor disability. They have poor score on cognitive, neuropsychological, educational and behavioral assessments even with no functional motor deficit; memory and attention or executive functions were impaired in severe groups. There is difficulty with language, spelling, letter recognition, visual and short term memory. ELBWchildren perform poorly on tests of visual-motor performance but are closer to controls on measures of language.

**Hearing impairment:**

Hypoxia damages the hair cells of the cochlea and hearing impairment is often due to hypoxia ischemia.
Seizures:
HIE grade III is associated with seizures and is a bad prognostic factor for long term intact neurology.

IVH, PVL:
Although incidence of periventricular hemorrhage has decreased, the rates of PVL remained unchanged thus leading to adverse neurological outcome. Isolated visual impairment can rarely be seen as a consequence of PVL.

RDS, BPD, PDA:
Complications of RDS like asphyxia, cardiac arrest affect neurodevelopmental outcome. RDS may be associated with PDA. Delayed closure of the PDA is associated with hypoxia, acidosis, increased pulmonary pressure secondary to vasoconstriction, systemic hypotension, immaturity, and local release of prostaglandins, which dilate ductus. There is a relationship between early adrenal insufficiency, ductal patency, airway inflammation and the development of BPD. VLBW infants with PDA are at increased risk of more prolonged and more severe RDS, bronchopulmonary dysplasia and death due to progressive respiratory failure. Although 85 to 90 % of all infants surviving RDS after requiring ventilatory support with respirators are normal, the outlook is much better for those weighing more than 1500 gm. The long term prognosis for normal pulmonary function in most infants surviving RDS is excellent. Survivors of severe neonatal respiratory may have significant pulmonary and neurodevelopmental impairment. Somehow pulmonary hemorrhage does not seem to be associated with increased long term morbidity.

NEC:
Premature infants with NEC who require surgical intervention or who have concomitant bacteremia are at increased risk for adverse growth and neurodevelopmental outcome.

Hyperbilirubinemia:
Disruption of the blood brain barrier by disease, asphyxia, and maturational changes in blood brain barrier permeability affect risk. The precise blood level above which free bilirubin is toxic for an individual infant is unpredictable, the duration of exposure needed to produce toxic effects is unknown. The more the preterm the infant the greater the susceptibility to kernicterus. The commonest outcome known is athetoid cerebral palsy.

Renal injury in asphyxiated newborn:
Oliguria in the perinatal period is a sensitive indicator of infants at risk for long term neurologic deficits. Oliguria was significantly associated with clinical signs of HIE, including seizures, death and long term neurological deficits.

Sepsis:
There is association between infection and brain injury, including severe
intraventricular hemorrhage and periventricular leukomalacia. This leads to poor neurodevelopmental outcome. Elevated peripheral neutrophil counts in the first 96 h of life in term infants with HIE may contribute to abnormal neurodevelopmental outcome. Neonatal infections among ELBW infants are associated with poor neurodevelopmental and growth outcomes in early childhood\textsuperscript{11}. Atonic cerebral palsy is caused by postnatal factors such as toxin, trauma, infection\textsuperscript{7}.

**Hypoglycemia:**
Both premature and full term infants are at risk for serious neurodevelopmental deficits from equally low glucose levels. The risk is related to the depth and duration of hypoglycemia. Early diagnosis and treatment of neonatal hypoglycemia is crucial to prevent future neurological sequelae. Even in the absence of gross hypoglycemic encephalopathy, it can be a cause of epilepsy even in full term babies due to delayed feeding which has been elegantly described by Udani et al. Aggressive correction of hypoglycemia is more important in presence of additional perinatal risk factors\textsuperscript{12,13}.

**Treatment Modalities in a High Risk Neonate**

**Surfactant treatment:**
In surfactant treated neonates moderate and severe pulmonary haemorrhage is associated with an increased risk of death and short term morbidity. Long term morbidity not affected.

**Role of ventilation:**
The prognosis of ELBW with ‘protected long term’ Ventilation remains grim. Those intubated have diminished survival and high rates of impairment. Parents of these infants should be informed of changes in prognosis as the time of ventilation increases. Severe hyperoxaemia and severe hypocapnia were associated with adverse outcome in infants with post-asphyxial HIE. During the first hours of life, oxygen supplementation and ventilation should be rigorously controlled\textsuperscript{14}.

Early HFOV when used with a lung recruitment strategy in combination with surfactant replacement may ameliorate acute neonatal lung injury that predisposes some preterm infants to develop chronic lung diseases\textsuperscript{15}.

**Role of inhaled nitric oxide:**
The results of NO inhalation are very promising. Use of NO has improved neurodevelopmental outcomes at two years of age. Inhaled nitric oxide decrease the risk of chronic lung disease and death, as well as of severe intraventricular hemorrhage or periventricular leukomalacia\textsuperscript{16}.

**Hyperoxia:**
Exposure to the extra uterine environment to high inspired oxygen concentrations produces cellular damage due to free radicle release and is a well known cause of retinopathy of.
prematurity (ROP), however lower the gestational age, lower the birth weight, and the sicker the infant add to the risk is for ROP10.

Environment, socioeconomic status, parental education and nutrition, as predictors of outcome:

While under nutrition is harmful, over nutrition seems to harm lower birth weight in adult life. Studies all over suggest, that environment certainly contributes in the long run which has been shown by Dr Chaudhari et al in her “Pune children Studies- Biology vs Environment”. Bavdekar et al from Pune have shown that a combination of small size at birth, followed by accelerated weight gain during childhood appeared to be responsible for an increased risk of insulin resistance in prepubertal children consistent with earlier findings by Barker et al. Combination of perinatal risk factors and non optimal rearing conditions can lead to poor developmental outcome, where as good rearing conditions can ameliorate the risk. Low social class is a frequently used index of non optimal rearing environment because of the associated social and economic disadvantages. Poor social class is powerful determinant of intellectual status in preterm infants.

Conclusion

Short term outcome and 2 years outcome are predicted by combination of BPD, PDA, IVH, PVL, RDS, NEC etc. The more the diagnoses an infant has the more severe those diagnoses the more likely that infant show adverse response on assessment at 18 to 24 months. Early outcomes do not reliably predict school age performance, behavioral problems, specific learning disabilities etc and studies have shown adverse outcomes have remained unchanged over last 2 decades. Therefore the need for a multidisciplinary follow up clinic under guidance of a neonatologist cannot be overemphasized which should be explained to the parents during discharge planning from NICU. Early interventions in such clinics are aimed at minimum disability and can rehabilitate the child but can not give cure. Therefore enhancing the recognition of high risk pregnancies, developing strategies to prevent preterm births, increasing maternal transports to level III prenatal centers, improving neonatal transport facilities to ensure prevention of hypoxemia and hypoglycemia and broadening the scope and availability of prenatal care to periphery will lead to better neonatal long term outcome. More attention may be paid to hypo/hyper oxemia and also hypocarbia and better infection control. Areas of research include usage of nitric oxide, optimal nutrition and early intervention programs for rehabilitation of children with adverse outcomes.
1. Wilson–Costello d. Is there evidence that long term outcomes have improved with intensive care?; Seminars in Fetal and Neonatal Medicine 2007; 12, 344-54.
Transient Tachypnea of Newborn: A Preventable Impediment for Neonatal Growth and Development

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Introduction
There are multiple factors responsible for the growth of human during neonatal period. This period is of immense importance with respect to the growth and development at later life. Neonatal growth is dependent on prenatal growth which in turn depends on maternal nutrition, maternal anthropometry, insulin and insulin like growth factor-2 and normal placental function. During postnatal period it is dependent on thyroid hormone, growth hormone, insulin like growth factor-1, nutritional factor, energy expenditure, neonatal care etc. Neonates are prone to different type of morbidities which influence their energy expenditure and growth. When babies are delivered normally at full term, there is very less risk of suffering from any unwanted phenomena. But nowadays there is an increasing trend of elective cesarean section; sometimes it is due to obstetrician’s decision and sometimes according to the choice of the prospective mothers and families. In many cases, this method of delivery can be avoided which can lead to smoother neonatal and infantile course. In this article, I want to report three full term neonates, delivered by elective cesarean section, had stormy neonatal life due to transient tachypnea of newborn (TTN). Normally this problem does not create much problem and lasts only for 48-72 hours, but that is not the universal rule and an unexpected disease course was experienced in these present cases. Moreover, in these cases, there was no other diagnosed genetic or maturational problem, birth asphyxia, metabolic disorder, maternal diabetes or any other ailments which can be blamed for giving rise to such circumstances. They were required to get admitted in neonatal intensive care unit (NICU) with symptoms of TTN with severe respiratory distress and needed to be mechanically ventilated. Mechanical ventilation itself is a risk factor for developmental delay and also other morbidities which independently hampers neonatal and infantile growth and development. So in these cases the unwanted outcome could be avoided just by allowing the babies to be delivered normally. But these cases can act as eye opener to learn to prevent such type of upshot.

Case Presentations
Case 1:
One male full term newborn was delivered by elective caesarean section and on the first day of life only, developed increasing respiratory insufficiency with
severe tachy-dyspnoea. He was shifted to NICU. After a brief treatment attempt with continuous positive airway pressure (CPAP), endotracheal intubation was imminent. On the second day of life his chest X-ray revealed a picture similar as respiratory distress syndrome grade II (with airbronchogram). The necessity of high pressure ventilation (PIP-30 mm hg) and poor oxygenation indicated the therapy with surfactant (curosurf @ 200 mg/kg). After the application of surfactant and high frequency oscillatory ventilation (HFOV) [Initial setting: Mean airway pressure 16, amplitude 28, frequency 10Hz, FiO2 40%] the respiratory situation improved gradually. On the 12th day of life he was extubated to CPAP but within 2 days he required to be reintubated as his requirement of FiO2 increased gradually and respiratory distress was visible. Blood gas was also showing acidotic picture. He was put under synchronized intermittent mandatory ventilation (SIMV) with mild setting and required to be mechanically ventilated 3 more days. Ultimately during the 3rd week of life he could be successfully extubated and the repeat chest X-ray showed a normalized view. On the 25th day of life he was discharged from NICU without any further problem.

**Case 2:**

The second baby was a full term female newborn presented with respiratory insufficiency shortly after birth. She was shifted to NICU and chest X-ray was done which showed a mediastinal and apical pneumothorax of the left lung and a picture similar to respiratory distress syndrome grade I. By applying CPAP her condition did not improve. Ultimately after endotracheal intubation, mechanical ventilation (SIMV) and application of surfactant the situation improved (initial setting PIP - 22, PEEP – 7, Flow 10, FiO2 30%, rate – 60/min, Ti – 0.33 sec., Te – 0.67 sec.). The pneumothorax was reabsorbed spontaneously. She could be extubated by 10th day of life. Later her clinical course was problem free.

**Case 3:**

This mature male newborn developed a severe respiratory distress syndrome directly after birth. He was shifted to NICU for respiratory assistance. Endotracheal intubation was necessary as he was non responsive to CPAP. After introduction of conventional mechanical ventilation (SIMV) his respiratory condition improved for a short period but deteriorated again demanding high frequency ventilation. The chest x-ray showed fluffy opacities all over the lungs. Some areas of lungs were hyper aerated with prominent vascular marking around the hila. Ultimately he improved with HFOV. On the 7th day of life he could be extubated without any further complication.

These three cases showed the example of extreme end of the picture of transient tachypnea of the newborn.
Discussion
TTN is also known as delayed clearance of fetal lung fluid. It was first described by Avery and co-workers in 1966. In this situation, lungs are filled with fluid (20-30ml/kg body weight) instead of air and this fluid is quite distinctive from amniotic fluid or plasma.

In order for the fetus to complete the transition from intrauterine to extrauterine life, the lungs must clear this fluid soon after birth. The process of clearing liquid from the lungs actually begins 2-3 days prior to birth with a decrease in the rate of secretion of fetal lung fluid. However, the lung-liquid begins to clear in earnest with the onset of labor. It was reported from previous study that nearly two third of the total clearance of liquid that occurs during transition from intrauterine to extrauterine life, occurs during labor. With the onset of labor, the pulmonary epithelium changes from a chloride secreting membrane to a sodium absorbing membrane, with reversal of the direction of flow of lung liquids. This change is an active metabolic process involving increased sodium potassium adenosine triphosphatase (Na+-K+-ATPase) activity in the epithelial cells and serves to drive liquid from the lung lumen into the interstitium and also mediated in large part by transepithelial sodium (Na) reabsorption through amiloride-sensitive Na channels in the alveolar epithelial cells, with some contribution from mechanical factors and Starling forces.

In addition, as lung liquid contains very little protein, oncotic pressure also favours the movement of water from the air space back into the interstitium and from there into the vascular compartment.

Greater likelihood of caesarean section has been associated with TTN. Along with that, risk of respiratory distress secondary to TTN, surfactant deficiency, and pulmonary hypertension is also increased. Previous reports showed that infants not exposed to vaginal compression, have excessively high volumes of interstitial and alveolar fluid for the first few hours, though thoracic volume remains within normal range; i.e. liquid volume is increased whereas gaseous component is decreased after caesarean section. So infants born without labor do not have the opportunity for early lung liquid clearance and begin their extrauterine life with excess water in their lungs.

Newborn babies with TTN often are hypoproteinemic and decreased plasma oncotic pressure may delay the direct absorption of water into the blood vessels. Babies with this diagnosis can have elevated pulmonary vascular pressure and ventricular dysfunction which again can raise central venous pressure and impair thoracic duct function and also the removal of interstitial water by lymphatics. Increase of pulmonary vascular pressure gives rise to a decrease in cardiac output. As the accumulated liquid compresses the
compliant airways and cause obstruction, the infants develop respiratory distress. In TTN, on chest X-ray, some parts of lungs show hyperaeration due to gas trapping as a result of airway obstruction. The babies develop hypoxia due to continued perfusion of poorly ventilated lung units; also hypercarbia develops from mechanical interference with alveolar ventilation and from central nervous system depression. Blood gas analysis reveals respiratory acidosis which supports the presence of hypercarbia.

Though generally this condition does not last for longer time, but in some cases respiratory assistance in the form of CPAP, SIMV or HFOV is required. Sometimes surfactant is also required to treat the emergency situation of respiratory distress. Though initially it was speculated that structural mutation of surfactant protein B (polymorphisms of intron 4 and heterozygous 121 ins 2 mutation) is associated with this condition, no such relation was found but relation with caesarean section and male infants were established. So, surfactant washout or surfactant inactivation is responsible for the respiratory distress and exogenous surfactant administration leads to a successful treatment outcome.

Some of the babies with this condition require mechanical ventilation. A number of respiratory and non respiratory complications stem out of this treatment modality. Among the respiratory complications, airway injury, chronic pulmonary disorders, air leak are noteworthy. Mechanical ventilation also can give rise to infection, feeding intolerance, intraventricular haemorrhage, developmental delay. It is postulated that standard technique of applying positive pressure ventilation may itself lead to impaired alveolar growth, although the effect is enhanced by concomitant respiratory distress. Again newborn infants’ energy expenditure increases greatly by different co morbidities and to some extent by mechanical ventilation. For normal growth, a full term infant’s requirement increases gradually till the end of the second week of life and reaches to an optimum level of 100-120 kcal/kg/day. With infection, increased respiratory work during distress and ventilation this requisite becomes several fold higher but at the same time these situations give rise to feed intolerance. So for appropriate nutritional delivery, central venous line placement becomes essential which again is one of the route causes of infection. So ultimately a vicious cycle develops.

Long term follow-up study showed learning and school performance problem, growth and neuro-developmental delay in those children who were under mechanical ventilation for long time or developed broncho-pulmonary-dysplasia. Delayed onset of regular respiration is also a risk factor for cerebral palsy, mortality and growth
delay\textsuperscript{20}. So it can be said that frequent caesarean section which are performed nowadays can give rise to such unwanted and preventable complications which can act as a hindrance for normal development of those term neonates.

To be more precise, caesarean section is often performed without any strict medical indication. This is a clear risk factor for development of transitory tachy-dyspnoea with respiratory distress in mature babies. Severe pneumothorax can also occur in those cases due to strong shear forces.

But all these ailments are preventable. So it needs to be emphasized, that the risks of caesarean section have to be carefully discussed with the parents and a stringent guideline for its real indications has to be defined. Probably this can prevent the NICU admission after caesarean section "without medical indication". This policy can improve the growth and development of normal full term neonates without posing unnecessary impediment.


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for

**Indian Journal of Growth, Development and Behavioral Pediatrics**

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Developmentally Supportive Care for Premature Baby

Santosh Nimbalkar

Although a baby may need to be in the hospital nursery, it is not always a very comfortable or a restful place to be. Lights are on 24 hrs a day, machinery, alarms, people, telephones and radios can create a lot of noise and that necessary medical procedures are often painful. Even routine caretaking activities can be tiring and disruptive.

Boston psychologist Dr Heidi Als and others have researched and developed ways of making nurseries and neonatal care more “baby friendly”. They have recommended changes in the ways nurseries operate to help lessen the negative effects the hospital care provides and minimize the stress babies experience. In addition they recommend that the care each baby receives be adjusted to best fit that child’s needs and coping abilities. This approach known as individualized developmental care (or formally, NIDCAP—Newborn Individualised Developmental Care and Assessment Program), is designed to provide an environment in which a premature development can continue as normally as possible despite his early birth.

Research into its effects has shown that babies who are cared for using the individualized developmental care approach have fewer medical complications, shorter stays in the hospital, better weight gain, and fewer days on respirators. There may be long term effects of this approach as well. Some of the early research has indicated that babies cared for with the NIDCAP approach may show more organized behavior and better development in their first year of life. As a result of this work, many nurseries have made modifications in the nursery environment and to the way in which they provide care. For eg, medical and caregiving procedures are often clustered so that the baby can sleep undisturbed for several hours at a time. In addition during invasive or uncomfortable procedures, various comforting methods may be used to help babies stay calm. These include holding a baby in a curled position with hands or swaddling giving the baby something to grasp, or a pacifier to suck. Nursery may have a staff member—usually a nurse who has been trained in NIDCAP method. She will observe your baby, help plan his care and advice you and the staff on the best ways of handling him.

As a parent you can provide comfort and support to your growing baby in a
number of ways. These may include making modifications to your baby’s surrounding to minimize stress from noise and lights, as well as learning how best to hold and interact with your baby as he grows and matures.

**Observe the baby’s environment and try to minimize unnecessary noise and light**

(i) There are a number of simple adjustments you can make in your baby’s surrounding to help reduce the amount of disruptive stimulation that he receives. Make sure your baby is shielded from light either by adjusting the amount of light shining directly on him or by putting a blanket or other covering over his incubator or bassinet. Always close the doors to his incubator quietly instead of snapping them shut.

(ii) If the nursery seems particularly noisy because of a radio playing or phones ringing, talk to the staff about your concerns. They may be able to make adjustments to lessen the noise or move your baby to a quieter location.

(iii) Keep voices low around your baby, particularly when he is sleeping or move away from his bedside for conversations.

(iv) If your baby’s bed is located in an area of the nursery where there is a great deal of activity or foot traffic, see that he be relocated to a quieter place.

(v) If your baby is on a respirator make sure that the water that accumulates in the tubing is emptied regularly.

**Hold the baby in a flexed position and provide boundaries around him while he sleeps**

Premies, like all newborn babies feel more secure when they are swaddled securely in a blanket with their legs hooked up, arms bent, and hands brought together in front of them. When they sleep, they prefer to be touching or lying against something and will move in the incubator until they are against the wall or the bottom of the enclosure. By positioning your baby in a curled position and providing boundaries for him when he sleeps, you are not only helping him feel calm and comfortable but also encourage the development of the curled up position that babies naturally assume in the womb. Premies with their lack of muscle strength, have a hard time maintaining this position by themselves and if left alone, will lie spread eagle with straight arms and legs on the relatively hard, flat surfaces of their nursery beds.

To provide comfort to your baby and support his physical development, try the following measures.

(i) Swaddle your baby in a blanket with his arms and legs bent and hands brought together in front of him or to his face.

(ii) When you hold your baby, keep him in a slightly curled position with his
legs tucked up and his hands brought forward in front of him.

(iii) Create a nest for your baby to sleep in. Wool blankets or cloth diapers and place them around your baby to help keep his legs tucked.

(iv) If your baby must sleep on his back, provide rolls along his sides to keep his arms bent and hands brought together in front of him and another roll under his knees to keep his legs tucked up.

(v) In the hospital, babies sometimes sleep on artificial lambskins or water beds to soften the surface on the bed. Artificial lambskins and water beds are recommended for use only in the hospital where your baby is continuously monitored. They shouldn’t be used at home as they are associated with higher risk of Sudden Infant Death Syndrome (SIDS).

Learn to read babies cues and pace your activities with him accordingly.

As discussed earlier in this chapter, premature babies tend to express themselves through physical changes and behavior. As you spend more time with your baby and as he matures you will begin to recognize how he signals that he is getting tired or upset and the things he does to calm himself. The following technique may help your child stay calm or regain equilibrium if he has become upset.

(i) Provide one form of stimulation at a time. If you rock him, don’t talk; if you feeding him, try not to look him in the eye, while your holding shield his eyes from strong light. Add more types of stimulation slowly, watching your baby for signs of stress.

(ii) When your baby signals that he is getting tired and needs some time out give him a rest period by cutting back on some of the stimulation he is receiving. For eg if you are rocking and looking at him, look away and just hold him quietly, perhaps shading his eyes from light until he relaxes again or decrease the intensity of the stimulation by talking softly, or rocking more slowly. If these approaches don’t work, your baby may simply need to be placed back in his incubator or bassinet to rest and sleep.

(iii) Help your child bring his hands to his face or mouth, or offer him your little finger or a pacifier to suck on.

(iv) Handle and move him slowly and gently.

(v) If your baby must be unwrapped from his blankets during certain procedures, use your hands to keep his arms and legs tucked and create boundaries around him. This will comfort him and help him feel more secure.

(vi) Apply gentle but firm pressure on your baby’s back or chest with your open hands. This will help him to
block out other stimulus, calm down and organize himself.

4. The preemie parents companion’. Madden SL.

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*IJGDBP*
Genetic Counseling in Autism

Monidipa Banerjee
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Abstract
With increasing incidence of autism (1 in 150), families with an affected member have a right to know regarding their risk of having another autistic child. 5-10% cases of autism have an identifiable medical disorder. Remaining 90% of cases of autism, there is no identifiable cause (idiopathic). This is a study through parental interviews, on the experiences of parents going for a second child, after having their first child diagnosed as autistic. The second part of the study, looks at the awareness among obstetricians when dealing with this population of patients.

The concerns of families need to be addressed by professionals with knowledge of autism. Since obstetricians and pediatricians are most likely to face these questions, there should be some guidelines on this issue.

Objective:
This is a study through parental interviews, on the experiences of parents going for a second child, after having their first child diagnosed as autistic. The idea is to bring out the awareness of genetic predisposition in autism, and availability of adequate counseling services.

The second part of the study looks at the awareness among obstetricians when dealing with this population of patients.

Method
14 families were identified for the interviews. Each of these families had gone for a second child after having their first child diagnosed as autistic. The profiles of the 14 families were as follows:

(i) Father:
Aged between 30 – 50 years, average age- 39.5 years.
*Occupation* – Service 8, law 1, doctor 2, business 3.
*Highest educational qualification* – 7 were graduates and 7 were post graduates.

(ii) Mother:
Aged between 25 – 45 years, average age- 35 years
*Occupation* – House Wife 9, law 1, doctor 1, service 1, teacher 2.
*Highest educational qualification* – 12 were graduates and 2 were post graduates.

(iii) Age of first child (autistic) when
second child was born:
3 to 8 years, (average age - 4.8 years)

(iv) **Average monthly family income:**
5 had a family income of 10,000 to 20,000/per month and 9 had a family income of >20,000/ per month.

(v) **Family:**
13 stayed in nuclear families and one belonged to an extended family.

(vi) **Place of residence:**
12 families were city based while 2 were from towns.

The following questions were discussed with either or both parents on a one to one basis by a single interviewer:

1. Why do/did you want a second child? (tick one or more)
   a. Want to experience a normal baby?
   b. Want a baby whatsoever.
   c. Somebody to look after the 1st child.
   d. others (specify)

2. What/who motivated you? (tick one or more)
   a. Family
   b. friend/peer
   c. Self Husband/Wife
   d. Unplanned pregnancy
   e. Professional

3. Why do you think it happened? (First child having autism)

4. Any history of similar problems in the family?

5. Did you know autism has a genetic predisposition?

6. Did you go for counseling before conceiving or giving birth to your second child?

7. If yes, to whom?

8. What were you told?

9. Did you tell your Obstetrician about your first child? What did he/she say?

10. What tests were done antenatally?

11. Did you think of the possibility that the second child may be autistic?

12. Did you want a boy or a girl second time? Why?

Father- age
occupation
highest educational qualification

Mother- age
occupation
highest educational qualification

Sibling – age sex
Average monthly family income
Family – nuclear/extended
Place of residence- city/town/village
contact no.
e-mail

The following data were obtained:

1. **Why did you want a second child?**

Four couples want a baby whatsoever, 5 couples wanted a sibling to look after the first child, and
in 5 cases the pregnancies were unplanned.

2. **What/ who motivated you?**
   Seven couples took the decision themselves, jointly. 5 pregnancies as mentioned were unplanned. 2 couples were influenced by professionals, into having a second child.

Further questionnaire

1. **Why did you want a second child?**
   Want a baby whatsoever - 4  
   Somebody to look after the first child - 5  
   Unplanned pregnancy - 5

2. **What/whom motivated you ?**
   Self H/W - 7  
   Unplanned pregnancy - 5  
   Professional - 2

3. **Why do you think it happened?**
   (1st child born with autism)
   Nine said Don’t know – 9(64%) while 5 (36%) had different thoughts of their own.
   The different explanations given were:
   Child fell & fainted at 1 yrs 3 months – 1; Child fell at 2 yrs – 1; Genetic predisposition – 1; Lack of oxygen at birth – 1; Multiple USG/ polyhydramnios - 1.

4. **Are there history of similar problems in the family ?**
   No - 12(85%) yes - 2(15%) (Border phenotye, MR)

5. **Did you know autism has a genetic predisposition ?**
   No - 11(78%). Yes - 3 (22%)

6. **Did you go for genetic counseling?**
   No – 11(78%), yes – 3(22%)

7. **To whom?**
   Of the 3 who went for genetic counseling, 2 went to the pediatrician, and 1 went to a referral institute.

8. **What were you told?**
   All 3 were advised that no antenatal tests were available, and they had a higher risk of having another child with autism than general population.

9. **Did you tell your Obstetrician about your first child?**
   Yes – 13(93%) no - 1

   What did he/she say?
   They received the following answers:
   don't know – 1  
   may not be repeated – 2  
   your decision – 4  
   continue your pregnancy – 2  
   did not comment – 2  
   leave it to God – 1  
   have to take a chance - 1

10. **What tests were done antenatally?**
    TORCH, Triple test, USG were done in 7 cases and USG only in the
other 7 cases.

11. **Have you thought of the possibility that the 2nd child may be autistic?**
   Yes - 13(93%). No 1(7%)

12. **Did you want a boy or a girl second time?**
   Anything - 13(93%). Girl - 1 (7%)

13. **Why a autism is less in girl?**

**Results:**
Summarizing the above data it was seen that,
(i) 36%(5) - wanted a 2nd child to look after the 1st child
(ii) 85%(12) – it was a Self decision by couples
(iii) 85%(12) – there was no family history
(iv) 29%(4) – Blamed autism on factors in and around birth
(v) 78%(11) - Did not know autism has a genetic predisposition
(vi) 22%(3) - Thought of genetic counseling before having the second child
(vii)93%(13)- Told their obstetricians about their first child
(viii)93%(13) - Thought the 2nd child could be autistic

**Method**
The second half of the study looked at awareness among obstetricians when dealing with this population of patients. 20 Obstetricians were identified, all of whom were postgraduates in their field. They were all city based practitioners. They were asked to fill up a questionnaire consisting of 5 questions.

**Questionnaire**
1. What is autism? (tick one or more)
   - Developmental delay
   - Psychiatric illness
   - Childhood schizophrenia
   - Neurodevelopmental disorder
   - Mental retardation
2. What causes autism? (tick one or more)
   - Hereditary disorder (Mendelian)
   - Chromosomal disorder
   - Genetic disorder Familial factors
   - Environmental factors
   - Multifactorial
3. Write 3 most important features of autism.
4. Incidence of autism in recent years?
5. A couple whose 1st child is autistic, comes to you for advice regarding second pregnancy
   What would you advice?

The following data were obtained:

1. **What is autism?**
   - Dev. delay - 6. Psychillness - 1
   - ND disorder - 10. MR 3
2. **What causes autism?**
   - Genetic disorder -2
3. Write 3 most important features of autism.
The following answers were obtained:
(i) Hyperactivity, attention deficit
(ii) Child can’t think, can’t mix with other children
(iii) Developmental problem, low IQ level
(iv) MR, motor dysfunction, coordination problem
(v) Delayed speech, irritable
(vi) Unsocial
(vii)6 wrote, no idea

4. Incidence of autism in recent years?
(i) Don’t know - 12
(ii) Increased - 4(20%)
(iii) Decreased - 3
(iv) 2-5%-1

5. A couple whose first child is autistic, comes to you for advice regarding second pregnancy. What would you advice?
(i) Counseling and further advice - 4 (20%)
(ii) Refer to a pediatrician – 2 (10%)
(iii) No idea – 6 (30%)
(iv) Chance of repeat minimal – 2 (10%)
(v) Go for 2nd pregnancy – 4 (20%)
(vi) Ask for Karyotyping of both parents -1 (5%)
(vii) Whole battery of tests including Chorionic Villus Sampling -1(5%)

Results
Summarizing the above data it was seen that,
(i) 50% thought autism was a neurodevelopmental disorder
(ii) 90% thought cause of autism to be multifactorial
(iii) Only 20% was aware of increased incidence of autism in recent years
(iv) 30% had no idea about autism
(v) There was no clear cut idea regarding advice for next pregnancy

Discussion
With increasing incidence of autism (1 in 150), families with an affected member have a right to know regarding their risk of having another child with autism. The concerns are not limited to parents only. It may extend to siblings and cousins of autistic individuals. With increasing awareness of the importance of genetic influences on autism, there is now a demand from families with an affected member for advice regarding their risk of having an autistic child.

5-10% cases of autism have an identifiable medical disorder. Remaining 90% of cases of autism, there is no identifiable cause (idiopathic). Inheritance is likely multifactorial (genes plus environment). Exciting progress is being made in the journey toward discovery of genes conferring risk for autism and autism spectrum disorders.
Current research evidence available gives families empirical recurrence risks only. There are no prenatal or carrier state diagnostic tests.

Genetic counseling is a consultative service. Before counseling parents it is important to know what parents’ expectations from the consultation are. It is important to first assess main concerns of parents. One also needs to assess their knowledge of diagnosis. This gives an idea as to what they hope to gain from the session.

The study shows that in 85% of cases there was no family history of autism. Almost 80% of the couples did not know that autism had a genetic predisposition, and 1/3rd blamed autism on factors in and around birth. Because of lack of awareness only 3 out of 14 thought of genetic counseling. However almost all of them confided in their obstetrician regarding their first child, and feared that the second child could be autistic as well.

The concerns of families requesting genetic counseling need to be addressed by professionals with knowledge of autism. There is a need to give information regarding recurrence risks not only for autism but also for the broader phenotype. In addition, couples may have other issues they wish to discuss, which may influence their reproductive decisions.

Geneticists are available in select centers only. Under these circumstances obstetricians and pediatricians are best placed to guide parents.

The study shows that obstetricians did not have clear guidelines regarding advice for next pregnancy. They also had limited knowledge on autism. Since obstetricians and pediatricians are most likely to face these questions, there should be some guidelines on this issue. Appropriate referrals should always be made wherever possible.

**Conclusion**

With increasing incidence of autism (1 in 150), families with an affected member have a right to know regarding their risk of having another autistic child. The concerns of families need to be addressed by professionals with knowledge of autism. Since obstetricians and pediatricians are most likely to face these questions, there should be some guidelines on this issue. Appropriate referrals should always be made wherever possible.

Breaking News

Jaydeep Choudhury
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That medicine is not just a science and its practice requires art and tact is most evident when a doctor has to break news. A bad news cannot be converted to good news. Knowledge of normal psychology will help inform and make the process easier. Specific skills can be learned and applied. Thus good communication is the key. But the foundation is empathy. Breaking news is not like giving an update, one must be focused and understand the situation from “inside”. The approach should be with caring. It is a complex communication task – requires expert verbal and non-verbal skills. Poor delivery can create serious miscommunications regarding prognosis of illness or purpose of care. The experience may stay in the mind long after the initial shock has been dealt with. The way a doctor or other health or social care professional delivers bad news places an indelible mark on the doctor/professional-patient relationship.

The person who delivers news may have to cope with some difficulties. Like the savior complex, where a physician feels that he is a savior and should always come out with positive results. Perspective of disability and limitations of the physician is another issue. One has to realize that certain situations are beyond control. Then there may be discomfort with negative emotions.

What is bad news?

Bad news can mean different things to different people. Bad news may be “any information, which adversely and seriously affects an individuals view of his or her future” or, in situations where there is either a feeling of no hope, a threat to a person's mental or physical well-being, risk of upsetting an established lifestyle, or where a message is given which conveys to an individual fewer choices in his or her life. The common denominator is that bad news is a message, which has the potential to shatter hopes and dreams leading to very different lifestyles and futures.

Withholding bad news from patients was commonly practiced until recently. Hippocrates advised Concealing most things from the patient while you are attending to him. Give necessary orders with cheerfulness and serenity crevealing nothing of the patients future or present condition. For many patients chave taken a turn for the worse by forecast of what is to come. In 1847, the American Medical Association first code of medical ethics stated, The life of a sick person can be shortened not
only by the acts, but also by the words or the manner of a physician. It is, therefore, a sacred duty to guard him carefully in this respect, and to avoid all things which have a tendency to discourage the patient and to depress his spirits. A 1961 survey of 193 physicians revealed that 169 (88%) routinely withheld cancer diagnoses. Furthermore, they often used euphemisms such as “growth” to describe cancer. The policy was “to tell as little as possible in the most general terms consistent with maintaining cooperation and treatment.” However, the same study found that most patients desired the truth regarding their diagnosis.

In recent decades, this model of patient care has been replaced by one that emphasizes patient autonomy and full disclosure. Honest disclosure of diagnoses, prognoses, and treatment options allows patients to make informed healthcare decisions that are consistent with their goals and values.\(^3\)\(^4\).

**Conveying Information to Patients**

One should not break news in a hurry. The following is the Girgis and Sanson-Fisher guidelines on conveying information to patients about serious disease or death.\(^5\)

1. Ensuring privacy,
2. Allowing adequate time,
3. Assessing patients' understanding,
4. Giving information about diagnosis and prognosis simply and honestly, avoiding euphemisms,
5. Encouraging patients to express feelings,
6. Being empathic,
7. Giving a broad but realistic time-frame concerning prognosis,
8. Arranging a review.

The bottomline is that parents want their doctor to be honest, compassionate, caring, hopeful and informative.

**Six Step Protocol for Breaking News**

Robert Buckman has outlined a six step protocol for breaking bad news.\(^6\) The steps are:

1. **Getting started:**
   The meeting should be held in a private setting with both physician and parent comfortably seated. It is helpful to start with some questions to indicate to the parent that this conversation will be a two-way affair.

2. **Finding out how much the patient knows:**
   By asking a question such as, “What have you already been told about the illness?” one can begin to understand what the parent has already been told, or how much the parent understood about what’s been said, the parents level of technical sophistication, and the parent's emotional state.

3. **Finding out how much the patient
wants to know:
It is useful to ask parents what level of detail they wish to cover. Parents want additional information regarding the diagnosis, chances of cure, the side effects of therapy and a realistic estimate of recovery.

4. Sharing the information:
Decide on the agenda before sitting down with the parents, so that one has the relevant information at hand. The topics to consider in planning an agenda are: diagnosis, treatment, prognosis, and support or coping. However, an appropriate agenda will usually focus on one or two topics. Long lectures are overwhelming and confusing. Medical terms should be translated into English.

5. Responding to the patients feelings:
If the parent’s reaction is not understood, it will leave a lot of unfinished business, and one will miss an opportunity to be a caring physician. Learning to identify and acknowledge a parent’s reaction is something that definitely improves with experience.

6. Planning and follow-through:
At this point one needs to synthesize the parent’s concerns and the medical issues into a concrete plan that can be carried out in the parent’s system of health care. Outline a step-by-step plan, explain it to the parent, and contract about the next step. Be explicit about your next contact with the parent or the fact that you won’t see the parent. Give the parent a phone number or a way to contact the relevant medical caregiver if something arises before the next planned contact.

Providing a Plan
Planning and follow-through is very important. The doctor should synthesize the parent’s concerns and the medical issues into a concrete plan that can be carried out. Patients who have a clear plan for the future are less likely to feel anxious and uncertain. It may be helpful if the patient has the option to speak to the professional delivering the bad news at a later stage.

Some phrases and questions that help in good conversation are:
(i) “I wish I had better news” (as opposed to “I’m sorry, I have bad news”),
(ii) “I admire your courage,”
(iii) “I will be here for you,”
(iv) “What gives you hope and strength?”

Some statements which may be avoided include the following:
(i) “It could be worse,”
(ii) “We all die,”
(iii) “I understand how you feel,”
(iv) “Nothing more can be done.”
Things go Wrong
Things go wrong when we try to escape, we react in anger or we dilute the situation.

We try to escape:
We try to escape by inappropriate delegation. The senior most member of the unit should break the news. Distraction, intellectualization, minimization and empty reassurance should be avoided.

React in anger:
Denial, idealization, rehearsal of the story by the parents, some “unreasonable” demands, anger, frustration and blame may be natural reactions of the parents. A physician should not react in anger under such circumstances.

Dilute the agenda:
It is not wise to dilute the agenda. It is improper to discuss about billing, practical arrangements or try to distract with trivial comments. After all a bad news is a bad news and it is a serious matter to the parents.

What is the importance of documentation?
It is important that accurate records are maintained of the conversation and the information and details exchanged. These will assist in the future care of the patient and enhance communication within the multidisciplinary team including the patient’s General Practitioner.

What if the parent starts to cry while breaking news?
It is always better to wait for the person to recover and stop crying. If it seems appropriate, one can acknowledge and suggest a temporary break till the parent recovers. Most parents are somewhat embarrassed if they begin to cry and will not continue for long. The doctor must not try to act as if tears are an emergency that must be stopped. He should not run out of the room. One should show the willingness to deal with anything that comes up7,8.

When another Doctor has Conveyed the Message in an Insensitive Way
Physician should try to understand the parents feeling and response, and analyze what went wrong with the previous encounter. One should not slander the insensitive caregiver.

What is the impact on you as a health care professional?
Breaking bad news can be extremely stressful for the doctor or professional involved. The evidence suggests that the bearer of bad news experiences strong emotions such as anxiety, a burden of responsibility for the news and fear of a negative response. This stress can result in a reluctance to deliver bad news9,10.


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