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General Medicine

A RARE CASE REPORT OF CONGENITAL BILATERAL CEREBELLAR HYPOPLASIA

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ABSTRACT Cerebellar hypoplasia (CH) is a neurological condition in which the cerebellum the part of the brain is smaller than usual or not completely developed. The etiological spectrum of CH is wide and includes both primary (malformative) and secondary (disruptive) conditions. Primary conditions include chromosomal aberrations (e.g., trisomy 13 and 18) metabolic disorders (e.g., molybdenum cofactor deficiency and adenylosuccinase deficiency), genetic syndromes (e.g., Ritscher.Schinzel, Joubert, and CHARGE syndrome- coloboma, heart defects, choanal atresia, growth retardation, genital abnormalities, and ear abnormalities. and brain malformations (primary posterior fossa malformations e.g., Dandy–Walker malformation.) or global brain malformations such as tubulinopathies. Secondary (disruptive) conditions include prenatal infections (e.g:cytomegalovirus), exposure to teratogens, and extreme prematurity. The distinction between malformations and disruptions is important for pathogenesis and genetic counselling.Neuroimaging provides key information to categorize CH based on the pattern of involvement: unilateral CH, CH with mainly vermis involvement, global CH with involvement of both vermis and hemispheres, and pontocerebellar hypoplasia. Here we report a rare case of congenital Bilateral cerebellar Hypoplasia Presenting with Bilateral cerebellar Symptoms.

KEYWORDS : cerebellum; hypoplasia.

INTRODUCTION

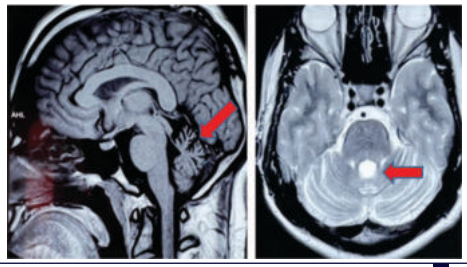
The Cerebellum derived from the Somatic afferent Portion of the Alar plate(rhombic lip) acts as monitor or modulator of Motor activity Originating in the other Brain centers.The Cerebellum is located in Posterior cranial fossa of skull i.e. dorsal to pons and medulla oblongata and separated from the occipital lobes by the tentorium cerebelli.The cerebellum controls the rate,direction,range and force of voluntary movements and through its vestibular connections ,corrects and adjusts the individual upright position in space. Thus Cardinal features of Cerebellar dysfunction involve disturbances in motor control, muscle tone regulation, and coordination of skilled movements .The First case of Cerebellar Hypoplasia was reported by French Neurologist Octave Crouzon in 1929. The Prevalence of Cerebellar Hypoplasia differed from time period .The Prevalence was 0.68 per 100,000 during 1997-2004, whereas Prevalence was 2.00 per 100,000 during 2005-2011.

Case Report

Here we had a 30 year old Male Right handed individual Presented to General Medicine OPD with Complaints of Slurring of Speech since Childhood. Patient attenders Noticed slurring of speech and frequent gaps between Syllables and there was undue Stress on the words Spoken by patient .However there was No reduction of word output during Conversation .He was obeying all commands told to Him .No History of Nasal Twang, Nasal Regurgitation of food .Patient Parents Gave history of Delayed Motor and social milestones .There was No neck holding and rolling over bed till 1 year of age. History of born out child from Second degree Consagnious marriage .No history of drug intake or infections during Pregnancy .Full term Normal delivery .Delayed cry noticed after 3min .However there was No history of NICU admission of the baby. Patient started walking after 3 years of age .History of Frequent falls Present .Parents used to support him while walking and Noticed Patient used to keep his legs wide apart .However by 5 years of age Patient was able to walk without falls. At the age of 6 years when patient started his schooling .Teachers told to Parents that your child was less interactive with others and he was poor in academics. However he completed his formal education upto Degree .At the age of 10 years .Patient Noticed involuntary movements in both hands during activity, specifically while reaching the objects.

At the age of 15 years Patient noticed Muscle fatigue in Upper limbs in the form of not able to lift heavy objects from the ground and Need of support of railings While climbing and getting down from stairs . However there is NO history of Further Progression and Improvement of disability over Past ten years .No History of seizures .No History suggestive of Cranial Nerve involvement, sensory symptoms and Bowel and Bladder involvement .NO History of Hypertension or diabetes .No History of any Neurological disorders in the family .CNS Examination : Right Handed individual .conscious ,oriented to time place person. Speech – Ataxic Dysarthria Present. Memory –Intact .Cranial Nerve examination-Horizontal Nystagmus Noted in Both eyes. Rest of Cranial Nerve examination –Normal. Motor System Examination: BULK –Upper limb and lower limb –NO significant wasting was noted .Tone –Upper limbs and lower limbs –Hypotonia Noted. Power –Upper limb and lower limb across all joints -5/5 noted .Reflexes –Superficial Reflexes. And Deep Tendon Reflexes are Normal, Bilateral Plantar –Flexor .Co-Ordination: Upper limbs –Finger Nose test –Past pointing Noted .Lower limbs –Knee heel test –Incoordination Noted .Intentional tremors Present .Rebound Phenomenon: present .Tandem walking –Incoordination Noted .Sensory system examination: Normal Gait: Wide Based gait noted. No evidence of truncal ataxia .NO signs of Autonomic Nervous System Disturbance noted .NO signs of meningeal irritation .Spine and skull: Normal. Probable Clinical diagnosis :30 year old male Presented with Chronic Symptoms of Dysarthria ,ataxia, Intentional tremors ,Hypotonia with delayed Physical and Motor Mile stones Suggestive of Bilateral cerebellar lesion of congenital etiology due to Bilateral cerebellar hypoplasia .

MRI Brain Done . It showed Reduced volume of Superior vermis with reduced volume of Superiomedial aspect of anterior lobe of cerebellum bilaterally.



DISCUSSION

The clinical phenotype associated with CH is wide and depends also on associated brain malformations or additional unrelated symptoms. Generally, children present with muscular hypotonia and global developmental delay and develop cerebellar signs later. Neurological findings include truncal ataxia (49–93%), hypotonia (47–49%), ocular movement disorders (40–46%), dysarthria (38%), intention tremor (9–35%).Intellectual disability is present in more than 60% of patients and is severe in 35% of them. Speech and language disorders are common and range from mild impairment to total absence of language development. We classify diseases with CH into four groups: (i) unilateral cerebellar hypoplasia, (ii) CH with mainly vermis involvement, (iii) global CH with involvement of both vermis and hemispheres and (iv) pontocerebellar hypoplasia. The Global Cerebellar Hypoplasia is associated with Genetic syndromes like Ritscher□Schinzel (3C) syndrome; Hoyeraal–Hreidarsson syndrome, neurofibromatosis type 1, Pallister–Killian syndrome. There is No Standard course of Treatment for cerebellar hypoplasia .Treatment depends upon the underlying disorder and the severity of symptoms .Generally ,Treatment is Symptomatic and Supportive .The Prognosis is dependent upon the underlying disorder .Some of the Disorders that are associated with cerebellar hypoplasia are Progressive ,which means the condition will worsen over time ,and will most likely have a poor prognosis .Other disorders that feature Cerebellar Hypoplasia are not progressive ,such as those that are the result of abnormal brain formation, during fetal development and might have better outcome. Here our case also have been diagnosed as Cerebellar Hypoplasia as result of abnormal brain formation during fetal development.

CONCLUSION

Cerebellar Hypoplasia is one of the rare cerebellar disorder of congenital etiology .Recent advances in neuroimaging and genetic testing have greatly improved the classification of CH disorders and identification of their causes. Many patients and their families now have access to diagnostic, carrier and prenatal testing. In addition, information on outcomes in patients with well□defined hindbrain malformations is continually improving. Identifying the genetic causes of these disorders is a major step toward understanding the underlying mechanisms that will be the best targets for specific therapies in the future.

Limitation of the Study: Because of Economical constraints of patient, Genetic studies have been not done.

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