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Familial Occurence of Moya Moya Disease in Three Sons of a Family- Rethinking its Genetics...

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I. Introduction:

Moyamoya Disease Is A Rare Progressive Vasoocclusive Disorder Of An Unknown Etiology. It Is Characterized By Progressive Stenosis Of Terminal Portions Of Internal Carotid Arteries Bilaterally, And The Main Trunks Of Anterior And Middle Cerebral Artery, And Is Associated With Collateral Vessels At The Base Of The Brain ('Moyamoya' Vessels).

The Term Moyamoya Is A Japanese Word Which Means "A Puff Of Smoke". It Was Coined By Suzuki And Takaku In 1969 To Describe The

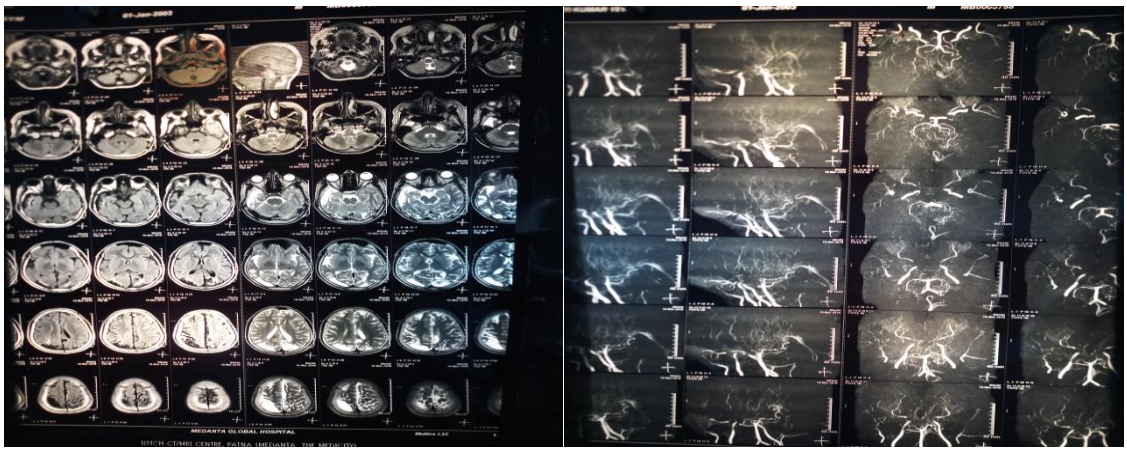
Angiographic Appearance Of The Collateral Vessels At The Base Of Brain In A Group Of 21 Patients With Internal Carotid Arterial Occlusion.

II. Case Report:

A Family With 3 Sons And 2 Daughters Were Evaluated For Moyamoya Disease. The Children Were Born At Term With Uneventful Neonatal Period And Normal Development History Upto The Age Of 1.5 Years, When The 3 Sons Got Their Symptoms Of Sudden Onset Weakness Of One Half Of The Body Along With Seizures. Daughters Of This Family Were Normal With No Such Complaints. Their Parents Were Also Evaluated For This, But Came Out To Be Normal. Later, Mr Angiography Was Done Which Revealed Presence Of Moyamoya Disease With Puff Of Smoke Appearance In All The Three Sons.

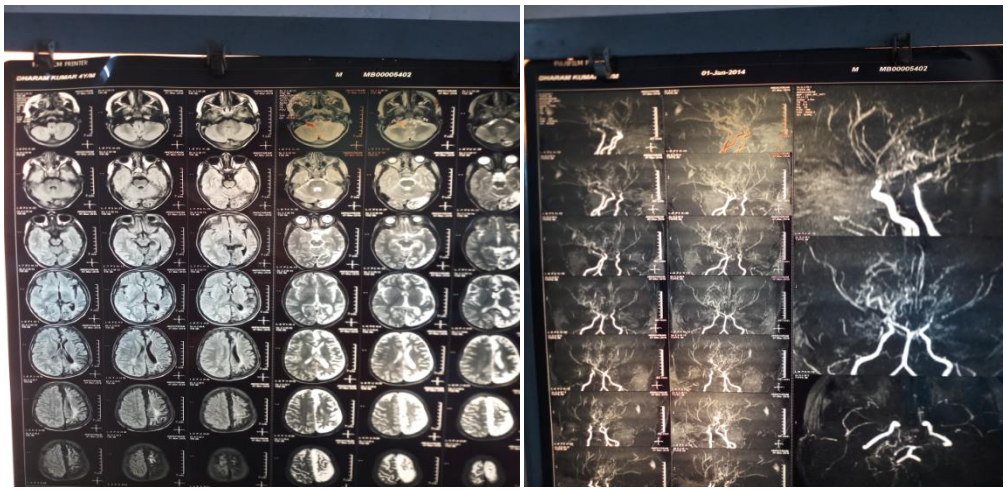
Case 1: 15 Yr Old Male Was Born By Normal Vaginal Delivery At Term, Achieved All Development Milestones Normally Upto 1.5 Yrs Age When He Suddenly Developed An Episode Of Generalized Tonic Clonic Convulsion, While Playing , Followed By Weakness Of Left Side Of Body Including Both Upper And Lower Limb And Was Unable To Walk Since Then. Gradually, Over The Years Weakness Of Lower Limb Improved But At Present He Was Unable To Hold Objects With His Left Hand. He Had No Seizures Since

Then. On Examination, Child Had Distal Weakness Of Left Upper Limb, With Bilateral Brisk Reflexes Suggesting Upper Motor Type Of Weakness With Same Side 7th Cranial Nerve Palsy Of Upper Motor Neuron Type. He Studied In Class 7th And Higher Mental Functions Had Subnormal Intelligence Not Upto His Age. Mr Angiography Was Done Which Revealed Occlusion Of Supraclinoid Segment Of Ica On Both Side With Marked Attenuation Of Both Aca And Multiple Small Collaterals Along The Circle Of Willis, Giving Puff Of Smoke Appearance- Suggestive Of Moyamoya Disease, With Gliosis In Right Fronto Temporal Region Suggesting An Old Infarct.



Case 2-7yr Old, 2nd Brother Had Similar History With Normal Birth And Development History And Had An Episode Of Seizure, Gtcs In Nature At The Age Of 1year After Which He Developed Sudden Onset Weakness Of Left Side Of Body Including Both Upper And Lower Limb. His Weakness Persisted In Both Upper And Lower Limb And He Had Multiple Episodes Of Weakness Since Then For Which He Had Taken Temporary Treatment For Those Episodes. On Examination He Had Upper Motor Type Of Weakness In Left Side Of Body With No Cranial Nerve Involvement, But His Higher Mental Functions Revealed Components Of Hyperactive Disorder, Otherwise Normal Functions. Mr Angiography Was Done Which Revealed Severe Stenosis Of Supraclinoid Segment Of Ica On Both Side With Multiple Collaterals Giving Puff Of Smoke Appearance Of Moyamoya Disease.

Case 3- 4 Yr Old, 3rd Brother, Again Had Similar Complaints Of Weakness Of Right Side Of Body Since The Age Of 1 Year Following An Episode Of Gtcs Convulsion , After Which He Had Multiple Episodes Of Convulsions Since Then, With Umn Type Of Weakness Of Right Side Of Body, With Normal Higher Mental Functions And No Cranial Nerve Involvement. His Mr Angiography Revealed, Occlusion Of Bilateral Ica , With Attenuation Of Mca And Aca With Multiple Collaterals Along The Circle Of Willis, Giving Puff Of Smoke Appearance Of Moyamoya Disease, With Diffuse Atrophy Of Left Cerebral Hemisphere And Mild Atrophy Of Right Fronto Parietal Lobe.



III. Discussion:

Both The Daughters Were Normal And The Parents Were Also Screened For This To Know About The Inheritance Of Moyamoya But To Our Surprise, Screening Mr Angiography Done, Of Both The Parents Came Out To Be Normal.

It Is Well Known That Moyamoya Is Familial In 10% Cases And Studies Have Revealed It As Autosomal Dominant Form Of Inheritance In Chromosome 17-Q25.3 With Involvement Of Rnf213 Gene. In Some Cases, X- Linkled Recessive Inheritance Is Also Seen If Mxm-4 Mutation Is Involved, Which Presents With Short Stature, Hypergonadotropichypogonadism With Facial Dysmorphism, Which Was Not In Our Case.

Involvement Of Three Members Of The Same Family, With Normal Parents And No Other Family History, Made Our Case Study Different Because, Autosomal Dominant Inheritance In This Case Can't Be Proved, Suggesting Some Other Mode Of Inheritance Or It Could Be Due To New Mutation In Dna Of Egg/Sperm In Which Parents Came Out To Be Normal , Or It May Be A Case Of Incomplete Penetrance With No Manifestations In Parents, Or With Variable Expression Or Somatic Mutations In Developing Embryo Which Need To Be Searched For And Further Evaluated For Better Knowledge Of This Rare Disease.

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