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LETTER TO THE EDITOR

Arthrogryposis Multiplex Congenita: Unthought of Silent Tragedy in Primipara

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Dear Editor,

In the Department of Obs/Gynae of KIST Medical College Teaching Hospital, Lalitpur, a 20 years Primigravida was booked having normal antenatal work up including normal anomaly scan except that she never perceived fetal movements that she complained in every regular antenatal visits. She was admitted multiple times in third trimester with same complaints but ultrasonogram and cardiotocograph (CTG) were normal all the times. At 38 weeks she developed pregnancy induced hypertension (PIH) and for the uncontrolled hypertension (AFI was normal) an Elective cesarean section was done with the difficult delivery of a baby which was impacted with increased tone of muscles, extended upper limbs and abducted lower limbs in typical fixed posture diagnosed clinically by the pediatrician as a case of Arthrogryposis multiplex congenita (AMC). The newborn had poor cry and low APGAR (female baby with birth weight 2.4 kg) and unfortunately died on the same day due to severe respiratory distress. This is a unique case we came across, which explains absent or reduced fetal movement despite normal CTG could be important predictor of AMC that one may not think of, can be alerted by abnormal fetal position devoid of intrauterine fetal movement that can be detected by 4D ultrasound which appears beneficial to be shared to bring awareness of this condition.

Described in 1841 by AE Otto and coined by WG Stern in 1923, Arthrogryposis multiplex congenita (AMC) derived from Greek words, arthon: "joints", gryposis: curvature "bent joints", is non-progressive congenital myodystrophy, characterized by crooked/contractures at two or more multiple joints (hands, feet, hips, knees, elbows, shoulders, wrists, fingers, toes, jaw and spine) often accompanied by fibrous ankylosis, an overgrowth or proliferation of tissue in the joint resulting in joint stiffness thereby predisposing to reduced /absent mobility.[1]AMC affects roughly 1 in every 3000-5000 live births without racial predilection with equal or double propensity for males; male to female ratio 2:1.[1] Possibly related to multiple factors {fetal neurological factors (Meningomyelocele, anencephaly, hydranencephaly, holoprosencephaly, spinal muscular atrophy, cerebro-oculofacial-skeletal syndrome, Marden-Walker syndrome); maternal factors(multiple sclerosis, diabetes mellitus, maternal infections, myasthenia gravis and hyperthermia) and external factors [oligohydraminos, intrauterine mechanical obstruction, trauma and fetal crowding (multiple pregnancy, uterine myomas)]}. Important ones being more than 35 specific genetic disorders mainly consisting of dystrophy, myopathy, myositis and mitochondrial dysfunctions which right away begins at first trimester of pregnancy.[2]

Unexplained fetal akinesia (absent/reduced fetal movement) for three weeks causes extra connective tissue to develop around the joint barring normal stretching of muscle/ tendons with reduced compliance of joints capsule and periarticular ligaments consequentially further aggravating fibrosis /contracture /fixation of the affected joints, earlier the occurrence graver the affection meaning that the joint development is embryologically normal condition arising mainly from immobility of joint, empirically essential for further normal joints development.

AMC are mainly of two types. 1. Amyoplasia (Classical Arthrogryposis) where the shoulders are internally rotated and adducted, the elbows are extended, wrists flexed and ulnarly deviated, fingers are stiff and thumb is positioned in the palm, hips are dislocated, knees may be extended, feets having severe equinovarus deformities, gastroschisis and bowel atresia affecting 10% majority (84%) with symmetric involvement of the upper and lower limbs. 2. Distal arthrogryposis are autosomal dominant disorders mainly involving the distal parts of the limbs, characterized by congenital contractures of two or more different body areas without a primary neurological and/or muscle disease.[3]

Other co-morbidities being cryptorchidism, respiratory problems due to thoracic deformity, neurogenic muscular atrophy, myopathy, congenital heart defect and dysphagia, latter causing aspiration was the single most important cause of death in approximately 20%. About 50% of infants that have contractures alongside a central nervous system dysfunction experience mortality in their first year of life. Hall reported mortality rate of 1% with limb involvement,

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7% with limb and other organ and 50% with limb and CNS involvement. Neonates who are ventilator dependent at birth obviously have poor prognosis.[3]

Myopathic or neuropathic disorders can be diagnosed by nerve conduction, electromyography and muscle biopsy. Had this baby of AMC in our case survived, we could have consulted orthopedic surgeon to plan for various corrective surgeries for better mobility and functionality. Orthopedic surgery is indicated for joint contractures resistant to stretching / splinting, others being osteotomy, tendon/muscle lengthening, tendon transfer for muscular limitation, tenotomy to release the joint from the pull of tendon and talectomy for equinovarus. Regarding the timing of surgery, soft tissue surgery such as bone and tendon transfers should be done around 3-12 months while osteotomies is done when the growth of the joint is near completed.[4] Proper physiotherapy, helps improve functionality correcting range of motion thus preventing further atrophy of muscles.[9]Specific management involves foot abduction braces, standing frames, stretching the hip flexors and prone positioning, thermoplastic serial splinting, training gross motor skills.

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