The Incidence of Orthopedic Related Problems in Lesch Nyhan Syndrome Patients and their Management

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ABSTRACT

Objective: To find out the incidence of orthopaedic related problems in Lesch Nyhan Syndrome patients and their management
Place and duration of study: The study was conducted at John Hopkins University from 1985 to 1993 during my post graduate training under supervision of Dr. Paul. D. Sponsceller Baltimore – USA
Patients & methods: Seven were included in this study whose diagnosis was confirmed by Bio Mechanical assay showing complete absence of HGPRT. Five out of seven patients had hips surgery along with Balancing of muscle forces by adductor and Psoase lengthening. Open reduction was performed in those hips where proper reduction was not possible by repositioning method. Femoral Osteotomy was only performed in patients older than five years and was fixed with blade plates. The orthopedic problems and there complication were studied.
Results: Neuromuscular hip dysplasia was present in majority of patients It was unilateral in two patients and bilateral in three patients. The mean age for diagnosis of hip dysplasia was eleven years. One patient had posterior subluxation of knee joint after septic arthritis of knee joint and was treated by antibiotic. Two patients had bilateral equinovarus deformity. Mild degree of scoliosis was noted in two patients and both were flexible and no surgical intervention was done.
Conclusion: The results of surgical interventions are satisfactory in our opinion and every effort should be taken to make the life of LNS patients comfortable, The caretaker should be educated properly as having pivot role and must be aware of incidence of re-fracture, postural adjustments, use of padding and prevention of bed sores
Key words: Lesh Nyhand Syndrome, femoral osteotomy, orthopaedic problems

INTRODUCTION

Lesch Nyhan Syndrome (LNS) is X linked disorder of purine metabolism first describe in 1964. This disease is characterized by Spasticity Chorioethetosis, Mental retardation and aggressive behavior including self mutilation. It is an X linked recessive disease caused by Hypoxanthine-Guanine-Phosphoribosyltransferase (HGPRT) deficiency. This leads to Hyper Urecemia, Hyperuricaciduria, and clinical manisfastation of Gout.

We reviewed 07 patients of Lesch Nyhan Syndrome (LNS). Results of orthopedic treatment and associated problems were recorded. Neuromuscular Hip were the most common problem noted. It was unilateral in two & bilateral in three patients.

The results of surgical procedures were satisfactory. Associated problems noted were fracture Autoamputation, infection, contracture, knee & ankle deformity along with scoliosis. We concluded that Lesch Nyhan Syndrome (LNS) patients can safely undergo surgical procedures and there results are similar to the patient with spastic cerebral palsy. The treating doctor should be aware of increased incidence of Heterophic Ossification as well as the incidence of implant failure and refracture in this disease if proper immobilization is not done.

MATERIAL & METHODS

All patients who presented at John Hopkins University from 1985 to 1993 were reviewed and only those patients (Seven) were included in this study whose diagnosis was confirmed by Bio Mechanical assay showing complete absence of HGPRT. Five out of seven patients had hips surgery along with Balancing of muscle forces by adductor and Psoase lengthening. Open reduction was performed in those hips

where proper reduction was not possible by repositioning method. Femoral Osteotomy was only performed in patients older than five years and was fixed with blade plates. The orthopedic problems and there complication were studied.

RESULTS

Hip Joint: Neuromuscular hip dysplasia was present in majority of patients It was unilateral in two patients and bilateral in three patients. Dysplastic acetabulum and coxa valga with femoral antivertion were found in three patients like patient in spastic cerebral palsy. The mean age for diagnosis of hip dysplasia was eleven years. Different surgical procedures like Adductor & Psoas tenotomy, Iliac Osteotomy, femoral varus de-rotational osteotomy, muscle lengthening and blade plate fixation was done according to the need and requirement of the patient. Complication noted in patient include loss of fixation (2), one broken and one cut out, infection, cast sore over anterior superior iliac spine and Heterotrophic ossification (three patients). We recommend as incidence of heterotrophic ossification is high so early measures should be taken to prevent it. More over if post operative immobilization after fixation is done than chances of complication are decreased.

Skeletal Problems in Lesch Nhyan Syndrom Patients

Age	Mental condition	Hip dysplasia (Rt Hip)	Hip dysplasia (Lt Hip)	Ankle deformity	Scoliosis	Fracture
18	mild	dislocated	normal	NAD	NAD	NAD
19	mild	normal	dislocated	NAD	NAD	Lt S/t # @ 11 yrs and Rt S/t # 15 yrs
23	Moderate	dislocated	Subluxated	Mild Valgus	15 degree TLdouble flexable	NAD
23	mild	Subluxated	Subluxated	NAD	NAD	NAD
14	Moderate	Subluxated	dislocated	Bilateral equinus	20 degree L-S flexable	NAD
27	Moderate	normal	normal	NAD	20 degree grade 1spondylolisthiasis	# Lt Radius
09	severe	normal	normal	NAD	NAD	NAD

Associated abnormalities noted in LNS patients

Infection	Renal stone	Gout	Seizures	Mutilation
Sup hip joint infection	+ ve	NAD	NAD	NAD
Septic arthritis right knee (haematogenus)	NAD	NAD	NAD	NAD
NAD	NAD	NAD	NAD	3 fingers amputated
NAD	+ ve	NAD	Present	NAD
Present	NAD	+ ve	NAD	Scared Hands
NAD	NAD	NAD	NAD	NAD
NAD	NAD	NAD	NAD	Lip mutilated

Knee joint: One patient had posterior subluxation of knee joint after septic arthritis of knee joint and was treated by antibiotic and pop cylinder cast for 6 weeks and other had geniu varus deformity which did not required any surgical treatment.

Ankle joint: Two patients had bilateral equinovarus deformity which was corrected with lengthening of tendon of Achllies (TAL) with satisfactory post operative result one patient had mild valgus deformity which does not require any surgical treatment.

Spine: Mild degree of scoliosis was noted in two patients and both were flexible and no surgical intervention was done. One patient had spondyliolesthiasis and also required no surgical intervention.

DISCUSSION

Although the characteristic physical finding of Lesch Nyhan Syndrome (LNS) is mental retardation, spasticity, chorioathetosis and self mutilation behavior and associated findings may be hyper uricemia, hyperuricaciduria along with clinical manifestation of gout and is caused by HGPRT deficiency but in 60% of patient with Lesch Nyhan Syndrome (LNS) delayed development finding was the first abnormality noted at the age of 6 month which on further investigation and observation lead to the diagnosis of Lesch Nyhan Syndrome (LNS). Self mutilating behavior starts at the age of 2 years and decreases with increase of age.

Self mutilating behavior is also present in patients with congenital insensitivity to pain but the difference between the two is that in congenital insensitivity to pain patients develops skin callous and in Lesch Nyhan Syndrome (LNS) there is loss of tissue. The exact cause is unknown but the lack of HGPRT activity in basal ganglia results in the super sensitivity of the dopamine receptors leading to self mutilating behavior. Although the aggressiveness decreases with the age but psychomimetic or serotonin precursor L-5 hydroxytryptophane is useful as self multilating agent. Precautionary measures such as extraction of primary teeth, mouth guard, elbow extension brace, may also prove useful in treating such patients.

Mental retardation is usually mild in these patients initially the child may loss ability to hold head and later on hypertonicity develops which leads to fracture and hip dysplasia. All patients on initial examination were diagnosed as having cerebral palsy but later on classical physical and clinical finding lead to the diagnosis of Lesch Nyhan Syndrome (LNS).

Pathological fracture are common in Lesch Nyhan Syndrome (LNS) and due to high muscle resting tune and spontaneous involuntary movements but the healing is much better in Lesch Nyhan Syndrome (LNS) as compared to the patients with spastic cerebral palsy.

CONCLUSION

1. The patients with LNS having orthopaedic problem can be treated safely similar to the patients with spastic cerebral palsy with satisfactory results.
2. Most common orthopaedic problem which needs surgical intervention in LNS is hip dysplasia
3. Hip fixation should be augmentated with spica cast in order to prevent implant failure or re-fracture.
4. Early measures should be taken to prevent heterotrophic ossification after surgery.
5. Once the patient is diagnosed as LNS early measures should be taken to prevent self mutilation
6. Other regional orthopedic problems like knee, ankle, and scoliosis, can be treated safely with conservative measures.
7. The results of surgical interventions are satisfactory in our opinion and every effort should be taken to make the life of LNS patients comfortable, The caretaker should be educated properly as having pivot role and must be aware of incidence of re-fracture, postural adjustments, use of padding and prevention of bed sores.

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