

# **MPS Type - VI / Maroteaux-Lamy Syndrome**

**(Mucopolysaccharidosis Type 6 / Lysosomal Storage Disorder)**

**Rare Genetic Disorder**

## **Prospective for HSCT**

**(Hematopoietic Stem Cell Transplantation)**

**Name of Child : AVISHI JAIN (Female)**

**DOB : 10-Feb-2012**

**Father's Name : Abhisek Jain / Mother's Name : Shikha Jain**

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**E-Mail : abhi99@gmail.com**

**Address : 193, Nemisagar Colony, Gate No. 3, Vaishalinagar,  
Queen's Road, Jaipur - 302021 (Rajasthan) INDIA.**

## **Medical History as on 25-August-2016**

### **Personal History:**

- Child's Name: Avishi Jain
- Gender: Female
- DOB: 10-Feb-2012 (Age: 4.5 yrs as on August '2016)
- Present Weight: 12.3 kgs
- Present Height: 88 cms.
- Age at which MPS VI Diagnosed: 2.5 yrs (Coarse facial features noticed)

### **Family History:**

- Father's Name: Abhisek Jain (DOB: 17-Sep-1982)
- Mother's Name: Shikha Jain (DOB: 20-Nov-1985)
- Younger Brother: Ayansh Jain (DOB: 17-April-2014) [healthy, negative for MPS VI]

### **Birth / Parents / Sibling:**

- Non-Consanguineous Parents
- She is First Child, Normal Delivery, Birth Weight: 3.2kg
- Second Child, Avishi's younger brother, is healthy, no MPS
- Brother Healthy, negative status of MPS VI confirmed by Enzyme Assay test

### **Present Condition:**

- Intelligent Child / Sharp Memory / Good Grasping
- Schooling since age 3 yrs / Amongst toppers in her class
- Walks normally, Runs, Jumps, Dances
- Normal speech and good comprehension & reasoning / Can talk properly in full sentences with fluency
- Liver Spleen normal, not enlarged
- Heart ok, confirmed by recent MUGA Scan
- No contractures
- Eye: Corneal clouding

**Present Condition (contd.):**

- Eye-Sight / Vision is normal, can recognize tiniest of objects and displays, can recognize all colours
- Dental Evaluation: OK

**Case for Stem Cell Transplantation:**

- No Government support or Insurance schemes in India to support Enzyme Replacement Therapy for MPS VI
- Wants to go for HSCT (Stem Cell Transplantation)
- HLA Tests done for self, brother, father, mother
- A, B and DRB1 values available for self, father & mother
- A, B values available For brother
- HLA match level with brother (HLA 3/4) [Brother's Umbilical Cord not preserved]
- HLA match level with father (5/6)
- HLA match level with mother (4/6)

**Visible MPS Symptoms at Present:**

- Bend in both wrists, slight curve in fingers, short stubby fingers
- Cloudy cornea and squint in left eye
- Enlarged Adenoids, sometimes breathes with mouth open while sleeping
- Concurrent running nose, resolved since Nov. 2015 after taking ongoing homeopathy medicine
- Short Neck
- Can raise her hands upwards 75% while changing clothes, cannot fully raise hands
- Gap between teeth
- Bone protruding behind shoulders
- Minute protruding bone on centre-top part of chest
- Stagnant weight since last one year : 12.5 kg to 12.2 kg
- Stagnant height since last one year, no increase in height, same clothes fit her

### **Diagnosis History of MPS VI:**

- **August 2014** - Coarse facial features observed by Paediatric Doctor at age 2 yrs 6 months.
- **October 2014** - MPS VI diagnosed at age of 2 yrs 7 months, by MPS Eltrophoresis Urine Test, in which abnormal bands in region of dermatan sulfate & heparan sulfate was noticed.
- **December 2014** - Further correlated by Enzyme Assay tests for MPS Type I, IVa, IVb and VI. MPS VI established by low value of ARSB enzyme.
- **January 2016** - MPS VI Gene Sequencing / Molecular Genetic Analysis for MPS VI done. N ARSB gene located on chromosome 5q11-q13 was sequenced. Mutation c.293T>G;p.L98R observed in homozygous condition.

### **DOWNLOAD LINKS of Medical Reports / History / Photos:**

**Request you to Download the following two ZIP Files which contain all medical history of my daughter:**

**1. Medical Reports of Avishi (24.8 MB zip file)**

[https://www.dropbox.com/s/42jw8e1gz9z3baj/Avishi\\_Jain\\_MPS\\_VI.zip?dl=0](https://www.dropbox.com/s/42jw8e1gz9z3baj/Avishi_Jain_MPS_VI.zip?dl=0)

**2. Images of CT-Scan Head (with DICOM software) (185.17 MB zip file)**

[https://www.dropbox.com/s/yo6pj4h3xnl873i/CT-Scan\\_NCCT\\_Head\\_Avishi\\_Jain.zip?dl=0](https://www.dropbox.com/s/yo6pj4h3xnl873i/CT-Scan_NCCT_Head_Avishi_Jain.zip?dl=0)

**Details of Test Reports / Medical Examination Records available in the above downloadable zip file-1:**

**Folder-1: MPS Test Reports**

1. Eltrophoresis Urine Test [25-Sep-2014]
2. Enzyme Assay tests for MPS Type I, IVa, IVb and VI [03-Dec-2014]
3. MPS VI Gene Sequencing / Molecular Genetic Analysis [09-Jan-2016]

**Folder-2: HLA Report (A, B, DRB1) & HLA Comparision [July '2016]**

1. HLA Report (patient, sibling, father, mother)
2. HLA comparision in spreadsheet ((ms-excel format & image format)

**Folder-3: Detailed Observations by Doctors**

1. 11-Oct-2014 : Genetics Doctor / Dr. I.C. Verma, Gangarams Hospital (Delhi)
2. 30-Nov-2014 : Paediatrician & Developmental Neurologist / Dr. S. Sitaraman (JK Lone Children's Hospital, Jaipur)
3. 10-June-2015 : Genetics Doctor / Dr. Ratna Puri, Gangarams Hospital (Delhi)
4. 07-June-2016 : Genetics Doctor / Dr. Madhulika Kabra, AIIMS (Delhi)

**Folder-4: Pre-HSCT Evaluation / Test Reports**

1. 24-June-2016 : List of Pre Transplant Evaluation Tests (Dr. Sameer Bakshi)
2. 16-July-2016 : Blood Test Reports (CBC, LFT, KFT, Viral Markers - HIV, HBsAg, Anti HCV) + Additional Vitamin D done
3. 02-July-2016 : MUGA Scan / Nuclear Heart Scan report
4. 14-July-2016 : GFR-DTPA / Nuclear Renal Scan (Kidney) report
5. 29-June-2016 : Prescription of Dental Evaluation
5. 29-June-2016 : Prescription of Ophthalmological Evaluation
6. 15-July-2016 : Prescription of ENT Evaluation + Audiometric Evaluation (Free Field Audiometry)

**Folder-5: CT-Scan of Head**

1. 30-June-2016 : Report of NCCT Head / CT-Scan Report
2. Link to download images/slides of CT-Scan / NCCT Head (with DICOM Viewer for Windows) given in a text file.

**Folder-6: Reports of other Tests/X-Rays done in past**

1. 01-Dec-2014 : X-Ray / Moderate Enlarged Adenoids.
2. 25-Apr-2014 : X-Ray Spine AP/Lat and X-Ray Both Wrist Joint AP (Bone Age)
3. 03-Oct-2014 : X-Ray of both Wrists AP/Lat
4. 04-Oct-2014 : USG of Abdomen
5. 03-Oct-2014 : Blood Test Reports (Haemogram, Calcium, S.Phosphorus, S. Alk. Phosphatase, Vitamin D)

**Folder-7: Photographs of Avishi in chronological sequence / age-wise**

1. 15 photographs in folder