حكومة دبي
GOVERNMENT OF DUBAI
LATIFA HOSPITAL

هيئة الصحة بدبي
DUBAI HEALTH AUTHORITY
مستشفى لطيفة

15 December 2016

## TO WHOM IT MAY CONCERN

## MEDICAL REPORT

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|---|---|
| **NAME OF THE PATIENT** | **: MAYED ALI MOHD A ALQASSAB** |
| **HEALTH CARD NO** | **: 2057 27 51** |
| **DATE OF BIRTH** | **: 02 MARCH 2004** |
| **NATIONALITY** | **: U.A.E.** |
| **GENDER** | **: Male** |

**Diagnosis:** Hereditary spastic paraplegia with 2 sibs similarly affected; a homozygous SPAST mutation has been identified on WES.

**Reference:**

- Medical report issued from Latifa hospital on 08 March 2016
- Medical report issued from Latifa Hospital on 07 November 2007, 25 June 2008 , 14 April 2009 , 11 March 2010 and 20 July 2014.
- Medical report issued from Germany on 09 March 2007.
- Medical report from Dr. Heakyung Kim, Visiting Consultant Pediatric Rehabilitation.
- Medical report on December 2006, 14 May 2007 and 11 December 2010 and 19 January 2012.
- Medical report from Dr Carlos De'sousa, Visiting Consultant Pediatric Neurology on 22 Nov 2010.

Mayed is a twelve years and 9 months old boy, known to have spastic and ataxic central motor defecits under follow-up in the Pediatric Neurology, Genetics and Orthopedic Surgery. He has been diagnosed to have hereditray spastic paraparesis. He has been seen by many visiting consultants and has also been seen in a hospital in Germany. He has had multiple sessions of botox injections here and elsewhere.

Mayed was born to consanguineous parents at term, there were no perinatal problems. He started walking late at twenty months and was found to have toe walking. Spastic lower limbs became obvious at that time.

As noted earlier, 2 of his sibs, Mirah and Mansoor, have similar problems; the three siblings are normal in their learning ability, but Mayed and his elder sister have experienced difficulty with handwriting and fine motor skills.

On examination, during the last visit in August 2016, Mayed looked alert and active, with normal growth parameters. He was attending a mainstrem school. His speech and handskills were near normal. He was ambulant with mixed spastic and ataxia of central origin. He showed scars of multiple tenotomies done earlier.



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He took a few steps without help, but he was ataxic with bilateral lower limb spasticity, and an exaggerated lumbar lordosis. A systemic review was otherwise unremarkable.

MRI done in September 2005 showed delayed white matter myelination, prominent extra- axial CSF spaces, sylvian of fissures and basal cistern with no mid line shift, well developed corpus callosum and no neuronal migrational abnormality.

He is under follow-up in the Rehabilitations unit and with Orthopedic Surgeon in addition to being seen by neurology team.

**Investigation:**

- Long Chain Fatty Acids: Test Result Units Essentially normal levels of VLCFAs with normal ratios. No indication of an inherited paroxysmal disorder. Plasma aminoacids chromatography. Aminoacid/acylcarnitin screen normal. Sugar aminoacid chromatography normal. Urine organic acid screening normal.
- Molecular cytogenetics - microarray test result: Normal
- MRI brain as noted above.
- Exome sequencing reports from genetic team notes:
  Heterozygous mutation (c.4579C>T, p.Gln1527* in LAMA1 gene associated with Poretti-Bolthauser syndrome.
  Hemizygous mutation UTR5 in APIS2 gene, x-linked recessive. It is a variant of uncertain clinical significance.

The genetic team has explained to the parents in detail regarding the genetic defect behind his neurological problems. The parents have been advised to continue regular, intensive rehabilitation program in the hospital and at home. He was advised to continue on vitamin D supplementation. He will be followed in the Genetics, Orthopedic Surgery and Pediatric Neurology Clinic. PT, OT, orthoses have to be continued as before. There is no specific treatment for his condition here or overseas.

This medical report is issued upon parent's request.

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