Suspected of MPS VI or Morquio A

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In my experience, the group of patient who usually come to receive treatments at Department of Orthopedics, QSNICH are the ones with disproportionate short stature, limb deformity and cord compression.

The main serious condition that needs correction is spinal cord compression that usually occurs at Occiput C1-C2 junction. But because of astonishment after seeing the outlandishness of the patient appearance, most doctors lost their focuses on the cord compression that leads to late diagnosis and ineffective treatment. Many doctors refuse to help because they suspect that the symptom caused by bad disease and impossible to cure. They inform the parents there is nothing they can do but accept things the way it is.

First case that will be presented is disproportionate short stature and limb deformity. The patient had walking difficulty and their ability to walk decreased within 1 year until they were unable to walk at all. However, there was no sign of mental disorder. From basic diagnosis found that they were in the type of Skeletal dysplasis: Spondyloepiphysseal dysplasia or MPS: Morquio syndrome. But what interested me was cord compression because these two groups usually had chances of cord compression at CVJ. In this case, pressing the cord at CVJ due to odontoid hypoplasia, C1-2 dislocation could be cured by reduction instrumentation and fusion. From the diagnosis with genetician, we suspected that patient had Spondyloepiphyseal dysplasia congenita because the result of urine screening for MPS was negative. However, blood enzyme testing hasn't yet been confirmed.

The second case is similar to the first one but from clinical characteristics and X-ray Bone Survey, the condition had been defined as MPS rather than Skeletal Dysplasia. There was TLC screening and Definite Diagnosis using blood enzyme testing from foreign lab and the expense was supported by private organization. There were odontoid anomalies but no sign of clinical cord compression. The case had been watched out for a while before the correction. However, MPS was beyond treatment. The patient was supported and encouraged mentally instead of having a real physical correction.

On the other hand, if the treatment had been taken seriously and the patient was given medication when they were young, it could reduce chances of spinal cord compression or abnormal weight bearing axis that could lead to joint pain or Osteoarthritis.

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ในการประชุม Asia Pacific Referral and Diagnosis Workshop focus on MPS VI and Morquio A
ซึ่งจัดขึ้นระหว่างวันที่ ๑๔ - ๑๕ กันยายน ๒๕๕๖ ณ เขตบริหารพิเศษฮ่องกง สาธารณรัฐประชาชนจีน