Brief Communication

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Mucolipidosis II/III in Malaysia

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Mucolipidosis (ML) is a very rare condition in Malaysia. During the period from 2003 to 2016, our department has diagnosed 12 patients (2 males and 10 females) with mucolipidosis. As shown in Table 1, ten patients have the more severe ML II (MIM #252500) phenotype. They have typical clinical features including coarse facial features, claw hands, dysotosis multiplex, hepatosplenomegaly, failure to thrive, recurrent respiratory infections and psychomotor retardation; clinically evident from early infancy. Most of the ML II patients died before 5 years old due to cardiorespiratory complications. Two patients have the milder ML III (MIM # 252600) phenotype. They have a later onset of symptoms and slower progression of disease. One of them

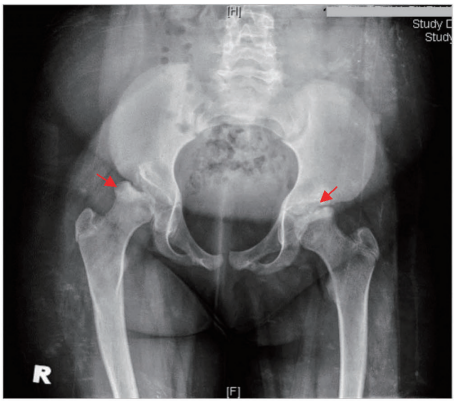

Fig. 1. X ray of patient 10: avascular necrosis of femoral heads (arrow).

Table 1. Summary of clinical characteristics of patients with mucolipidosis II and III included in the present study

Patient	ML type	Sex	Ethnicty	Parental consanguinity	Age at diagnosis (years)	Age at death (years)
1	II	Female	Malay	No	1 year	Alive at 4 years
2	II	Male	Malay	No	3 years 1 month	<5 years
3	II	Female	Malay	No	3 months	<5 years
4	II	Female	Malay	No	6 months	<5 years
5	II	Female	Malay	No	1 year 8 months	<5 years
6	II	Female	Chinese	No	2 years	<5 years
7	II	Female	Malay	No	1 year 5 months	<5 years
8	II	Female	Chinese	No	1 year	5 years 5 months
9	II	Male	Indonesian	Unknown	9 months	Alive at 6 years 9 months
10	III	Female	Indian	Yes	12 years	Alive
11	III	Female	Malay	No	10 years	Alive
12	II	Female	Malay	No	2 years 6 months	Alive at 3 years

*Classification of the type of mucolipidosis was assigned by each patient's treating physician.

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presented with pain and contracture of the joints of the hands at 10 years old, mimicking juvenile chronic arthritis. Another patient presented at 12 years old with bilateral hip joint disease, initially misdiagnosed as Legg-Calve-Perthes disease. Her pelvic and proximal femurs X ray showed avascular necrosis of femoral heads (Figure 1). Both ML III patients exhibited normal cognition and attended regular school. Biochemically, all patients have increased lysosomal hydrolases enzymes activities in plasma but low activities in cultured fibroblasts. Details about their genotypes were not available at the moment.