Information from medical charts	
Age at symptom onset mean (median; min, max) months	
All n = 18	77.1 (42.0 ; 0, 540)
Without outlier n = 16	53.0 (42.0; 3.0, 192.0)
Age at first consultation mean (median; min, max) months	
All n = 18	78.9 (42.0; 4.5, 540.0)
Without outlier n = 16	54.8 (42.0; 12.0, 192.0)
Age at diagnosis mean (median; min, max) months	
All n = 18	113.8 (60.0; 7.0, 540.0)
Without outlier n = 16	93.9 (60.0; 14.0, 324.0)
Diagnoses identified for study inclusion	**N (%)**
Australia	7 (39%)
South Korea	4 (22%)
Taiwan	3 (17%)
Japan	2 (11%)
Thailand	1 (6%)
Hong Kong	1 (6%)
Gender	**N (%)**
Female	14 (78%)
Male	4 (22%)
Pre-diagnostic key symptoms and findings	**N (%)**
Short stature	13 (72%)
Pectus carinatum	9 (50%)
Genu valgum	8 (44%)
Spinal abnormalities	7 (39%)
Gibbus/kyphosis	5 (28%)
Scoliosis	3 (17%)
Atlantoaxial instability	3 (17%)
Hip dysplasia	4 (22%)
Impacted joint range of motion	2 (11%)
Joint pain	2 (11%)
Advanced bone age	2 (11%)
Dermal melanocytosis	2 (11%)
Spinal cord compression	2 (11%)
Overgrowth within one year of age (“big baby”)	2 (11%)

- Other skeletal abnormalities experienced by patients included arachnodactyly shortened distal phalanges, dysplastic and fragmented proximal femoral epiphysis, flattening of femoral heads, acetabular irregularities, joint space narrowing
- Other general symptoms experienced by patients included upper respiratory infections, hernia repair, unsettled behavior

Impediments to diagnosis	N (%)
Atypical symptoms	5 (28%)
Subtle symptoms	4 (22%)
Symptoms associated with other diseases	4 (22%)
Marginally elevated or normal uGAG levels	4 (22%)
Delayed clinical suspicion	2 (11%)
Other radiologic differentials (pseudoachondroplasia)	1 (6%)
Misdiagnosis and other clinical diagnoses prior to Morquio A diagnosis	**N (%)**
Spondyloepiphyseal Dysplasias	5 (28%)
Gibbus/kyphosis	5 (28%)
Scoliosis	3 (17%)
Craniosynostosis	2 (11%)
Advanced bone/dental age	2 (11%)
Overgrowth	2 (11%)
Genu valgum	2 (11%)
Corneal opacity	2 (11%)
Cardiac conduction abnormality	2 (11%)
Legg-Calvé-Perthes Disease	1 (6%)
Leri-Weill syndrome	1 (6%)
Marfan syndrome	1 (6%)
Sotos syndrome	1 (6%)
Pseudoachondroplasia	1 (6%)
Truncal hypotonia	1 (6%)
Hypertolorism	1 (6%)
Splenomegaly	1 (6%)
Growth hormone deficiency	1 (6%)
Torticollis	1 (6%)
Sleep obstruction (snoring)	1 (6%)
Cervical lymphadenopathy	1 (6%)
Apnea/hypopnoea	1 (6%)
Truncal hypotonia	1 (6%)
Rheumatoid arthritis	1 (6%)
Harrison sulcus	1 (6%)
Lumbar lordosis	1 (6%)
Reversed Madelung deformity	1 (6%)
Autism	1 (6%)
Specialist responsible for Morquio A diagnosis	**N (%)**
Geneticist	10 (56%)
Radiologist	5 (28%)
Pediatrician	2 (11%)
Endocrinologist	1 (6%)