



**Fig. S1.** (a) X-rays of the patient's hands showed pronounced osteolysis of the middle and distal phalanges. (b) Chest x-rays showed thoracic scoliosis. (c) Three-dimensional reconstruction of the skull showed an open of cranial suture. (d) Cranial MRI showed leukoencephalopathy, hydrocephalus, and a widened subarachnoid space. (e) Cranial MRI showed distinctive cranial deformities, arachnoid cysts, mega cisterna magna, and cerebellar atrophy.