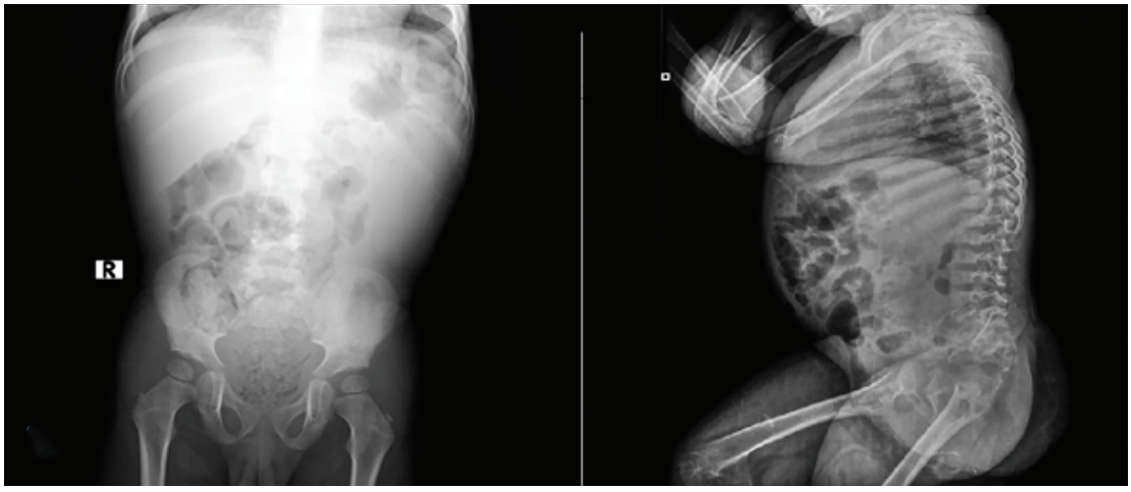
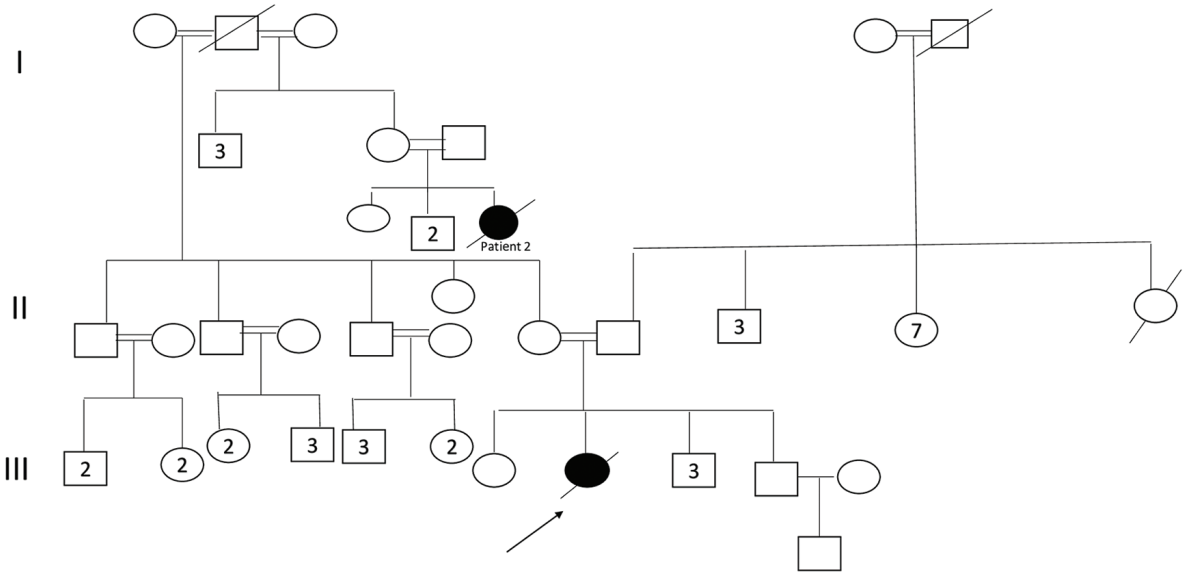
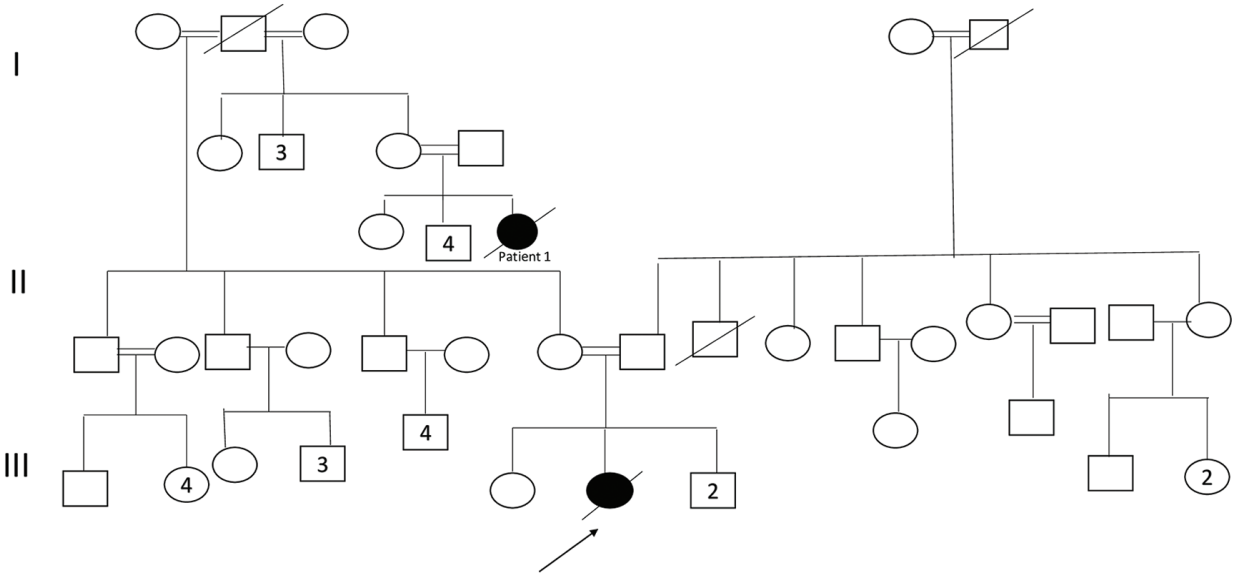


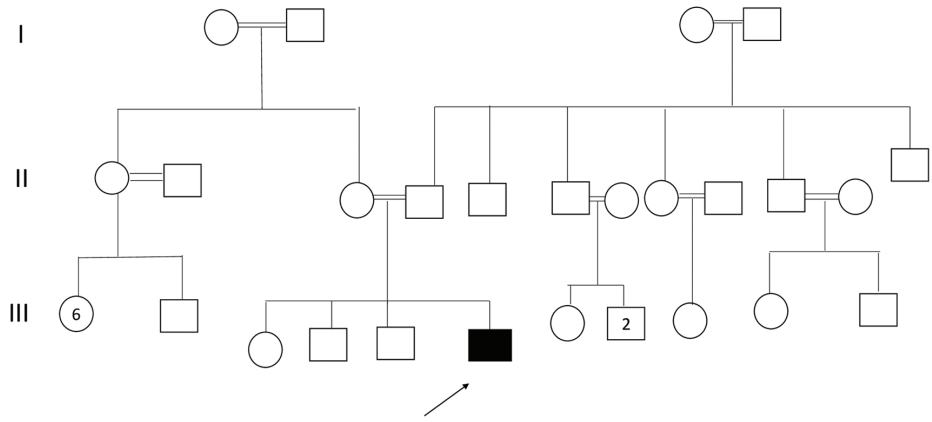
SUPPLEMENTARY FIG. 1. (a) Relvis X-ray of patient 2 demonstrating a hxpoplastic pelvis with shallew acetabular fossae and dysplastic femoral beads. (b) Lateral relvis view of patient 4 showing flattened vertebral bodies and abnormal spinal alignment.



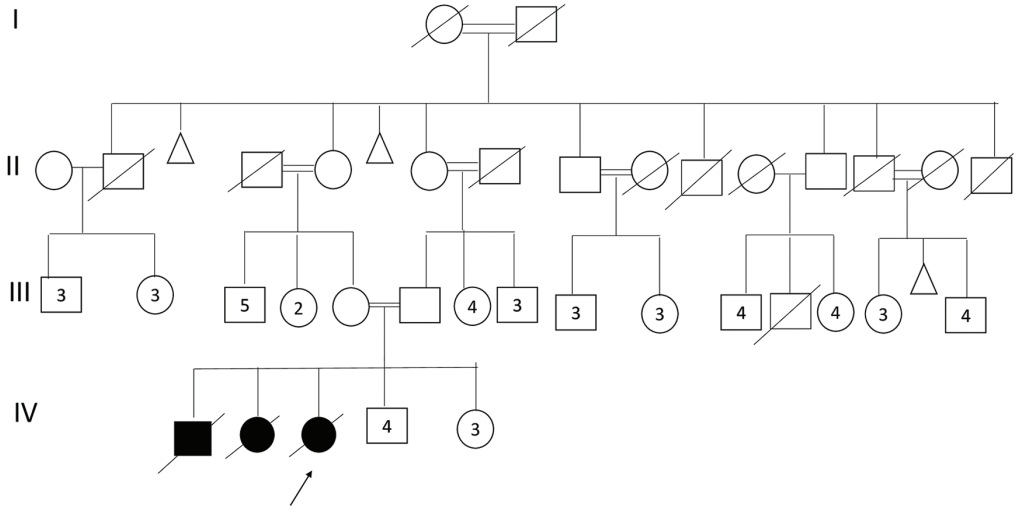
SUPPLEMENTARY FIG. 2. Pedigree of patient 1.



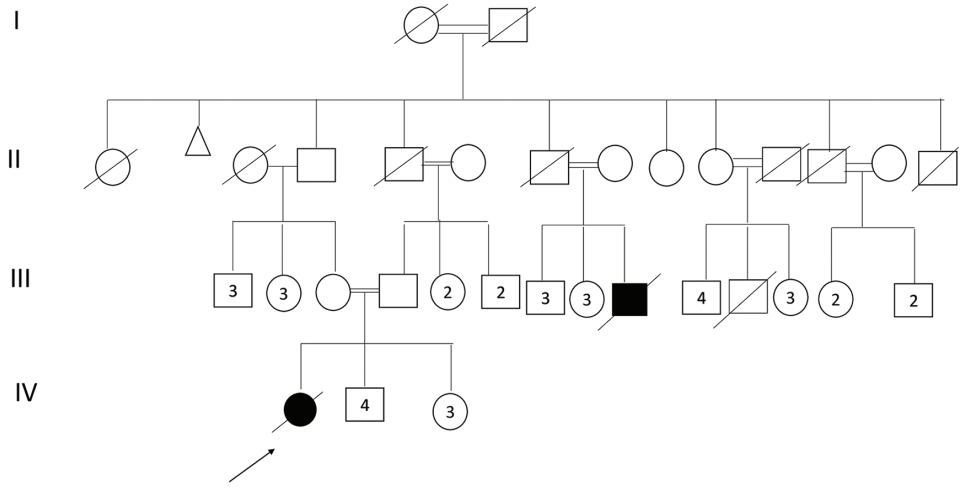
SUPPLEMENTARY FIG. 3. Pedigree of patient 2.



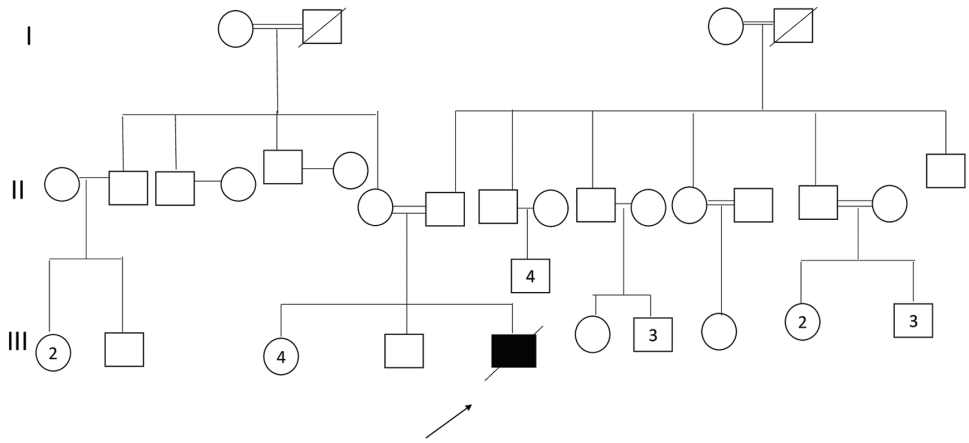
SUPPLEMENTARY FIG. 4. Pedigree of patient 3.



SUPPLEMENTARY FIG. 5. Pedigree of patient 4.



SUPPLEMENTARY FIG. 6. Pedigree of patient 5.



SUPPLEMENTARY FIG. 7. Pedigree of patient 6.

SUPPLEMENTARY TABLE 1. Comparison of Clinical Findings in the Current SIOD Cohort with Previously Reported *SMARCAL1*-Related Phenotypes.

Clinical feature	Present study (n = 6)	Previously reported SMARCAL1-related SIOD phenotype*
Growth retardation/short stature	All patients had significant short stature and low height SDS values.	Very common; proportion > 90% in most cohorts.
Dysmorphic facial features	Typical SIOD facies present in the majority of patients.	Characteristic facial dysmorphism frequently reported.
Spondylo-epiphyseal dysplasia (SED)	Radiographic SED findings documented in all evaluated patients.	Core skeletal manifestation in classic SIOD.
Nephrotic syndrome/proteinuria	NS or heavy proteinuria present in most patients at diagnosis.	Common; often steroid-resistant nephrotic syndrome.
Progressive kidney dysfunction/ ESRD	Four patients progressed to ESRD; one had advanced CKD stage IV.	High risk of early-onset CKD/ESRD widely reported.
Histopathology (FSGS/other)	FSGS in ¾ biopsied patients; C1q nephropathy in one.	FSGS is the predominant lesion; other patterns less frequent.
T-cell immunodeficiency/ lymphopenia	Lymphopenia and T-cell abnormalities present in several patients.	Typical; cellular immunodeficiency is a key feature.
Hypogammaglobulinemia	Observed in 2 patients with persistent proteinuria.	Reported in a subset of cases, often protein loss–related.
Recurrent/severe infections	Severe infections contributed to morbidity and mortality in multiple patients.	Frequently described due to immüne dysfunction.
Neurovascular events	Ischemic stroke and/or transient neurological events in multiple patients.	Well-documented association with cerebral ischemia and vasculopathy.
Autoimmune manifestations/ cytopenias	Autoimmune hemolytic anemia and bone marrow failure seen in 2 patients.	Autoimmune cytopenias reported in some cohorts.
Vascular complications (arteriosclerosis)	Arteriosclerotic changes noted in selected patients.	Described as part of SIOD vasculopathy spectrum.
Congenital renal anomalies (ectopic kidney)	Ectopic pelvic kidney present in one patient.	Rarely reported; contributes to expanding renal phenotype.
Novel *SMARCAL1* variant (c.1177C > T, p.Arg393*)	One patient with severe multisystem involvement and poor outcome.	Not previously reported; expands mutational spectrum.
Consanguinity	All patients born to consanguineous parents.	Frequently reported in SIOD, especially in high-consanguinity populations.

SIOD, Schimke immuno-osseous dysplasia; NS, nephrotic syndrome; ESRD, end-stage renal disease; CKD, chronic kidney disease; FSGS, focal segmental glomerulosclerosis.
*Data summarized from previously published SIOD cohorts and reviews including Boerkoel et al.,[16] Elizondo et al.,[3] Lücke et al.[17]