Title: Transition care for young people with rare bone and mineral conditions – a scoping review

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Table S1: Overview of the terms included in the literature search and the searches conducted in the databases

Documentation of literature search

Research question: Transition care for young persons with rare bone conditions – a scoping review

All searches were done 01 August 2025, by TMH, senior librarian at the University of Oslo, Medical Library at Ullevål Hospital, Research Skills and Literature Search Section, Oslo, Norway

Search syntax:

Ovid-databases	
exp/	Exploded index term
/	After an index term indicates a subject heading were selected.
.ti,ab,kf.	Search for a term in title, abstract and author keywords
.kw.	= keyword heading
*	At the end of a term indicates that this term has been truncated, diet* retrieves both diet, diets, dietary.
adj3	Search for two terms next to each other, in any order, up to 3 words in between.
Scopus	
TITLE-ABS-KEY	Search for word in title, abstract or keyword
AUTHKEY	Search for word in author's keyword
W/3	Search for two terms next to each other, in any order, up to 3 words in between.
PRE/3	Terms must appear in a specific order between words, where first term precedes the next within three words

Ovid MEDLINE(R)

#	Searches	Results
1	Bone Diseases/	23522
2	exp Rare Diseases/ and (bone or skeletal).ti,ab,kf.	762
3	((rare or genetic* or inherit* or heredit* or familial) adj2 (skeletal or skeleton or bone or mineral*) adj2 (disease* or disorder* or dysplas* or abnorm* or anomal* or deform* or syndrome* or malalign* or condition*)).ti,ab,kf.	2171

4	RBMC*.ti,ab,kf.	96
5	((bone or skeletal) adj2 (dysplasia* or disease* or disorder* or aplasia* or hypoplasia*)).ti,ab,kf.	49918
6	exp Osteogenesis Imperfecta/	5479
7	(Lobstein* adj (syndrome or disease)).ti,ab,kf.	103
8	(porak and durante).ti,ab,kf.	17
9	(osteogenesis imperfecta or achondrogenesis or Bruck syndrome or brittle bone or fibrogenesis imperfecta ossium or osteopsathyrosis or periostal aplasia or (Cole-Carpenter adj (dysplasia or syndrome))).ti,ab,kf.	6557
10	exp Achondroplasia/	2637
11	(achondroplasia* or achondroplasy or achondrosis or hypochondroplasia* or pseudoachondroplasia* or saddan or saddans or skeleton skin brain syndrome*).ti,ab,kf.	2646
12	exp Familial Hypophosphatemic Rickets/	980
13	(x adj2 linked adj2 hypophosphat*).ti,ab,kf.	1363
14	(XLH or rickets or osteomalacia or renal osteodystroph* or ckd-mbd).ti,ab,kf.	17070
15	(chronic kidney adj disease adj mineral adj bone disorder).ti,ab,kf.	233
16	exp Dysostoses/	20998
17	(dyostoses or dyostosis).ti,ab,kf.	3
18	exp Hypophosphatasia/	1142
19	(hypophosphatasia* or phosphoethanolaminuria or hypophosphatasis).ti,ab,kf.	1375
20	(phosphatase adj2 deficien*).ti,ab,kf.	423
21	(Rathbun adj2 (disease or syndrome)).ti,ab,kf.	3
22	exp Exostoses, Multiple Hereditary/	1435
23	(multiple adj2 (osteochondroma* or exostos*)).ti,ab,kf.	1558
24	(chondrodysplasia* or osteochondromatosis or ecchondrosis ossificans or exostotic dysplas* or familial exostos*).ti,ab,kf.	2797
25	(diaphyseal adj2 aclas*).ti,ab,kf.	56
26	(Bessel-Hagen adj2 (disease or syndrome)).ti,ab,kf.	9
27	exp Dwarfism/ or exp Dwarfism, Pituitary/	18282
28	(dwarfism or nanism or short stature or thanatophoric).ti,ab,kf.	21195
29	((growth hormone or gh or hgh) adj2 (insensitiv* or deficien* or defect* or resistan*)).ti,ab,kf.	12055
30	(((mulibrey or muscle-liver-brain-eye or pituitary) adj nanism) or perheentupa syndrome*).ti,ab,kf.	117
31	(marfan* adj syndrome*).ti,ab,kf.	7815
32	(marfan* adj2 disorder*).ti,ab,kf.	117

33	exp Arachnodactyly/	547
34	arachnodactyl*.ti,ab,kf.	902
35	(Loeys-Dietz adj2 syndrome*).ti,ab,kf.	848
36	(marchesani adj2 syndrome*).ti,ab,kf.	214
37	(congenital mesodermal dysmorphodystroph* or gemss or spherophakia-brachymorphia syndrome* or craniometaphyseal dysplasia*).ti,ab,kf.	181
38	(((Robinow* or Laron* or Larsen* or Kabuki* or Noonan* or cockayne* or progeria-like) adj syndrome*) or acromesomelic or progeroid nanism or omodysplasia* or (mesomelic adj4 dysplasia*)).ti,ab,kf.	5948
39	(congenital hypothyroidism or cretinism).ti,ab,kf.	5218
40	exp Campomelic Dysplasia/	83
41	((campomelic adj3 (dysplasia* or syndrome*)) or osteochondroma or cartilaginous exostos* or chondrosteoma*).ti,ab,kf.	3157
42	exp Osteochondromatosis/	1566
43	(osteochondromatosis or osteochondromatoses).ti,ab,kf.	670
44	exp osteosclerosis/ or exp melorheostosis/ or exp osteopetrosis/ or exp osteopoikilosis/ or exp Pycnodysostosis/	6006
45	(osteosclerosis or osteoscleroses or osteosclerotic or melorheostosis or melorheostoses or osteopetrosis or osteopetroses or osteopoikilosis or albers schoenberg disease or marble bone* or Raine dysplasia or dysosteosclerosis or pycnodysostos*).ti,ab,kf.	6604
46	exp Hyperostosis, Cortical, Congenital/	677
47	(hyperostosis or hyperostoses or hyperostostic or DISH or Forestier's disease or ((Caffey* or Forestier*) adj2 disease)).ti,ab,kf.	13361
48	exp Osteochondrodysplasias/ or exp Langer-Giedion Syndrome/ or exp Kashin-Beck Disease/ or exp Platybasia/ or exp Hyperostosis/ or exp Enchondromatosis/	39642
49	(osteochondrodysplasia* or dyschondroplasia* or dyschondrosteos* or chondrodystroph* or spondylodysplast* or spondyloepiphyseal dysplasia* or (spondylo-epi* and dysplasia*) or spondyloepiphyseal dysplasia congenita or SEDC or SEMD or (Kniest adj (dysplasia or syndrome*)) or Melnick-Needles syndrome* or mucopolysaccharidos* or MPS IV or eccentroosteochondrodysplasia* or eccentro-osteochondrodysplasia* or (morquio* adj (disease or syndrome*)) or galns deficien* or galactosamine-6-sulfatase deficien* or hypochondrogenesis or fibrochondrogenesis or achondrogenesis or atelosteogenesis or diastrophic dysplasia or otospondylomegaepiphyseal dysplasia or dyssegmental dysplasia or Schneckenbecken or opsismodysplasia* or bent bone dysplasia* or Stuve-Wiedemann or ((conradi and hunermann) adj2 syndrome) or Greenberg dysplasia or acrodysplasia* or ((giedion and langer) adj syndrome) or tricho rhino	19805

	phalangeal syndrome or trichorhinophalangeal syndrome or kashin beck disease or platybasia* or bone hypertroph* or hyperostosis or hyperostoses or (ollier* adj2 disease) or enchondrom* or chondroplasia angiomatosis or chondrodysplasia*).ti,ab,kf.	
50	(otopalatodigital adj2 (spectrum or syndrome or disorder*)).ti,ab,kf.	59
51	(chondrodystroph* adj2 myoton*).ti,ab,kf.	32
52	(melnick adj2 needles adj (syndrome* or osteodysplasty)).ti,ab,kf.	98
53	(endosteal hyperostosis or hyperphosphatasemia tarda or ((schwartz-jampel* or sja) adj2 syndrome*) or x-linked sed* or sed tarda or multiple epiphyseal dysplasia* or hyperostosis or myotonic myopath* or van buchem disease or sclerosteosis or osteoporosis pseudoglioma or OPPG syndrome* or eosinophilic granuloma* or (engelmann* adj (disease or syndrome*))).ti,ab,kf.	10629
54	exp Ellis-Van Creveld Syndrome/	565
55	(ellis van creveld adj2 (syndrome or dysplasia)).ti,ab,kf.	474
56	((chondroectodermal or mesoectodermal) adj2 dysplasia*).ti,ab,kf.	141
57	exp Chondrodysplasia Punctata/	1056
58	(chondrodysplasia or brachytelephalangic or ((conradi and hunermann) adj syndrome*)).ti,ab,kf.	1912
59	(CHILD syndrome or congenital hemidysplasia or fibula hemimelia* or (genu adj (valgum or varum or varus))).ti,ab,kf.	1972
60	exp Slipped Capital Femoral Epiphyses/	612
61	((coxa adj (magna* or valga* or valgus or vara)) or slipped capital femoral epiphys*).ti,ab,kf.	3032
62	exp Synostosis/	12465
63	(synostosis or synostoses or spondylocarp*).ti,ab,kf.	3857
64	exp Acro-Osteolysis/	318
65	exp Hajdu-Cheney Syndrome/	140
66	(cheney syndrome or acroosteolysis or acro-osteolysis or serpentine fibula).ti,ab,kf.	715
67	((hajdu and cheney) adj syndrome).ti,ab,kf.	190
68	exp Pseudohypoparathyroidism/	1781
69	(pseudohypoparathyroidism* or pseudohypoparathyreoidism* or (albright adj2 osteodystroph*) or phd ib or phd1b or php ia or phpia).ti,ab,kf.	1758
70	(hypoparathyreoidism or hypoparathyroidism).ti,ab,kf.	7293
71	exp Osteitis Deformans/	5331
72	osteitis deformans.ti,ab,kf.	901
73	((paget* adj disease) and bone).ti,ab,kf.	3811
74	((methotrexate or MTX) adj osteopathy).ti,ab,kf.	32
75	exp Myositis Ossificans/	2208

76	exp Osteolysis, Essential/	903
77	(fibrodysplasia ossificans or myositis ossificans or ossifying myositis or ossifying fibrodysplasia or stone man syndrome* or osteoarthropathy or paraosteoarthropathy or (Gorham-Stout adj (disease or syndrome)) or vanishing bone disease or melorheostos*).ti,ab,kf.	6141
78	exp "Fibrous Dysplasia of Bone"/	5334
79	((Albright* or (FD adj MAS)) adj3 (disease or syndrome* or spectrum or orosteodystrophia*)).ti,ab,kf.	1451
80	(fibrous dysplasia* or osteodysplasia* or osteodysplastic or fibrous osteodystrophy or osteodystrophia fibrosa or fibrodysplasia* or osteocraniostenos* or Desbuquois dysplasia*).ti,ab,kf.	6376
81	exp Osteitis Fibrosa Cystica/	1671
82	(osteitis fibrosa cystica or osteodystrophia fibrosa or bone fibrous dysplasia or dysplasia fibrosa or (Recklinghausen* adj disease)).ti,ab,kf.	3029
83	(fibrous bone adj (defect* or disease* or dysplasia*)).ti,ab,kf.	49
84	exp Leg Length Inequality/	3479
85	(leg length inequalit* or femoral deficiency or congenital femoral deficien* or proximal focal femoral deficien*).ti,ab,kf.	547
86	(limb* adj3 (reduction adj2 (defect* or deform* or deficien*))).ti,ab,kf.	450
87	(arachnodactyl* or brachydactyl* or ectromelia* or amelia* or hemimelia* or phocomelia* or sirenomel* or (proteus adj2 syndrome)).ti,ab,kf.	5430
88	exp Basal Cell Nevus Syndrome/	1469
89	(basal cell nevus syndrome or basal cell carcinoma syndrome or nbccs or fifth phacomatos* or (gorlin adj2 syndrome)).ti,ab,kf.	2196
90	exp Camurati-Engelmann Syndrome/	346
91	(engelmann* adj (disease or syndrome)).ti,ab,kf.	295
92	(chondroectodermal dysplasia* or diaphyseal dysplasia* or diaphyseal hyperostos* or metaphyseal dysplasia* or Cartilage-hair hypoplasia*).ti,ab,kf.	906
93	or/1-92	251415
94	Child/ or infant/ or adolescent/ or pediatrics/ or minors/ or (pediatric* or paediatric* or child* or infan* or toddler* or pubescen* or teen* or adolesc* or youth or (young adj (person* or patient*))).ti,ab,kf.	4684136
95	"Congenital, Hereditary, and Neonatal Diseases and Abnormalities"/	891
96	Child Guidance/ or Child Health Services/ or Child Guidance Clinics/ or Adolescent Health Services/ or Child Health Services/ or Adolescent Health Services/	27698
97	or/94-96	4685698
98	Adult/ or (adult or adults or adulthood or maturity or mature or grown-up* or "coming of age" or lifespan or life span or lifetime or lifelong).ti,ab,kf.	7235583

99	Disease Management/ or Personal Autonomy/ or Relational Autonomy/ or Patient Advocacy/ or Patient Participation/ or Self-Management/ or Self Efficacy/ or Self-Concept/ or Decision Making/ or Decision Making, Shared/ or Choice Behavior/ or Health Literacy/ or Adaptation, Psychological/ or Coping Skills/	394017
100	((patient* or self* or personal) adj7 (autonom* or manag* or admin* or advoca* or engag* or involv* or participat* or activ* or proactiv* or cope or copes or coping or sufficien* or sustain* or relian* or empower* or litera* or efficac* or mastery or organiz* or organis* or adapt* or navigat* or agency or choice* or choos* or determin* or decis* or decid* or independ* or depend* or control* or passiv* or prefer* or govern* or judg* or authorit* or unsupervis* or ((deal* or liv*) adj (with or through)))).ti,ab,kf.	2650454
101	or/98-100	9062602
102	97 and 101	2328769
103	Disease Progression/	205622
104	((clinical or disease* or disorder* or illness* or condition*) adj2 (course* or progress* or process* or develop* or evolv* or exacerbat* or deterior* or trajector*)).ti,ab,kf.	592573
105	Longitudinal Studies/	187111
106	(longitud* or long term or longterm).ti,kf.	396354
107	Follow-Up Studies/	716817
108	((follow up or followup) adj (study or studies)).ti,ab,kf.	63897
109	Socioeconomic Factors/ or Vulnerable populations/ or poverty/ or social class/ or Social Support/ or Financial Support/ or Family Relations/ or Health Behavior/ or "Social Determinants of Health"/ or Health Status Disparities/ or Social Problems/ or Happiness/	431154
110	(socioeconomic* or SES or SEP or sociodemographic* or socio-demographic* or SDOH or income or wealth* or poverty or educational level or level of education or educational attainment or well educated or better educated or academic achievement* or unemploy* or home owner* or tenure or affluen* or well off or better off or worse off or ethnic* or race or racial* or racis* or equit* or inequit* or inequalit* or disparit* or equality).ti,ab,kf.	1042639
111	((social* or socio-economic or socioeconomic or economic or structural or cultural) adj3 (advantag* or disadvantag* or exclude* or exclusion or include* or inclusion or status or position* or gradient* or hierarch* or class* or determinant*)).ti,ab,kf.	194824
112	"Quality of Life"/	308552
113	("quality of life" or life quality or qol or hrqol or "hr-qol" or hrql or ((patient* or personal*) adj2 (satisf* or happiness))).ti,ab,kf.	546781

114	or/102-113	5374659
115	"Continuity of Patient Care"/	21446
116	(continu* adj2 (care or healthcare)).ti,ab,kf.	29465
117	Transition to Adult Care/	2576
118	(transition care or healthcare transition or transition clinic* or "transfer of care").ti,ab,kf.	1852
119	((transit* or transfer*) adj3 (adult* or care or healthcare or clinic* or program* or care or process* or period or plan* or strateg* or model* or phase or tool* or guideline* or recommendat* or standardi* or journey* or trajector*)).ti,kf.	35683
120	((transition* or transfer) adj2 (program* or care or process* or period or plan* or strateg* or model* or phase or tool* or guideline* or recommendat* or standardi* or journey* or trajector*)).ti,ab,kf.	109313
121	"Continuity of Patient Care"/st [Standards]	2605
122	exp Transition to Adult Care/st [Standards]	285
123	exp Adolescent Health Services/st [Standards]	837
124	exp Child Health Services/st [Standards]	2860
125	exp Adolescent Medicine/st [Standards]	117
126	(transition* or transfer* or continu*).ti.	444680
127	or/115-126	555912
128	93 and 114 and 127	449
132	limit 128 to (danish or english or french or german or norwegian or portuguese or spanish or swedish)	399

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#	Searches	Results
1	bone disease/	38963
2	exp rare disease/ and (bone or skeletal).ti,ab,kf.	4161
3	((rare or genetic* or inherit* or heredit* or familial) adj2 (skeletal or skeleton or bone) adj2 (disease* or disorder* or dysplas* or abnorm* or anomal* or deform* or syndrome* or malalign* or condition*)).ti,ab,kf.	3228
4	((bone or skeletal) adj2 (dysplasia* or disease* or disorder* or aplasia* or hypoplasia*)).ti,ab,kf.	73410
5	osteogenesis imperfecta/	10705
6	(Lobstein* adj (syndrome or disease)).ti,ab,kf.	177
7	(porak and durante).ti,ab,kf.	39
8	(osteogenesis imperfecta or achondrogenesis or Bruck syndrome or brittle bone or fibrogenesis imperfecta ossium or osteopsathyrosis or	9508

	periostal aplasia or (Cole-Carpenter adj (dysplasia or syndrome))).ti,ab,kf.	
9	achondroplasia/	4681
10	(achondroplasia* or achondroplasy or achondrosis or hypochondroplasia* or pseudoachondroplasia* or saddan or saddans or skeleton skin brain syndrome*).ti,kf.	2575
11	familial hypophosphatemic rickets/	404
12	(x adj2 linked adj2 hypophosphat*).ti,ab,kf.	2259
13	(XLH or rickets or osteomalacia or renal osteodystroph* or ckd-mbd).ti,kf.	14405
14	(chronic kidney adj disease adj mineral adj bone disorder).ti,ab,kf.	352
15	dysostosis/	2430
16	(dyostoses or dyostosis).ti,ab,kf.	9
17	hypophosphatasia/	2718
18	(hypophosphatasia* or phosphoethanolaminuria or hypophosphatasis).ti,ab,kf.	2354
19	(phosphatase adj2 deficien*).ti,ab,kf.	598
20	(Rathbun adj2 (disease or syndrome)).ti,ab,kf.	4
21	hereditary multiple exostosis/	1786
22	(multiple adj2 (osteochondroma* or exostos*)).ti,ab,kf.	1939
23	(chondrodysplasia* or osteochondromatosis or ecchondrosis ossificans or exostotic dysplas* or familial exostos*).ti,ab,kf.	3739
24	(diaphyseal adj2 aclas*).ti,ab,kf.	72
25	(Bessel-Hagen adj2 (disease or syndrome)).ti,ab,kf.	15
26	thanatophoric dwarfism/ or dwarfism/ or short limbed dwarfism/ or pituitary dwarfism/	12057
27	(dwarfism or nanism or short stature or thanatophoric).ti,ab,kf.	32427
28	((growth hormone or gh or hgh) adj2 (insensitiv* or deficien* or defect* or resistan*)).ti,ab,kf.	17946
29	(((mulibrey or muscle-liver-brain-eye or pituitary) adj nanism) or perheentupa syndrome*).ti,ab,kf.	148
30	(marfan* adj syndrome*).ti,ab,kf.	11234
31	(marfan* adj2 disorder*).ti,ab,kf.	194
32	arachnodactyly/	1539
33	arachnodactyl*.ti,ab,kf.	1048
34	(Loeys-Dietz adj2 syndrome*).ti,ab,kf.	1234
35	(marchesani adj2 syndrome*).ti,ab,kf.	298

36	(congenital mesodermal dysmorphodystroph* or gemss or spherophakia-brachymorphia syndrome* or craniometaphyseal dysplasia*).ti,ab,kf.	266
37	(((Robinow* or Laron* or Larsen* or Kabuki* or Noonan* or cockayne* or progeria-like) adj syndrome*) or acromesomelic or progeroid nanism or omodysplasia* or (mesomelic adj4 dysplasia*)).ti,kf.	5444
38	(congenital hypothyroidism or cretinism).ti,ab,kf.	7597
39	campomelic dysplasia/	344
40	((campomelic adj3 (dysplasia* or syndrome*)) or osteochondroma or cartilaginous exostos* or chondrosteoma*).ti,ab,kf.	3891
41	osteochondroma/	4929
42	osteochondroma*.ti,ab,kf.	4685
43	osteosclerosis/ or melorheostosis/ or osteopoikilosis/ or Albers Schoenberg disease/ or pycnodysostosis/	13874
44	(osteosclerosis or osteoscleroses or osteosclerotic or melorheostosis or melorheostoses or osteopetrosis or osteopetroses or osteopoikilosis or (albers schoenberg adj (disease or syndrome)) or marble bone* or Raine dysplasia or dysosteosclerosis or pycnodysostos*).ti,ab,kf.	9480
45	Caffey disease/	899
46	(hyperostosis or hyperostoses or hyperostostic or DISH or Forestier's disease or ((Caffey* or Forestier*) adj2 disease)).ti,kf.	5345
47	chondrodysplasia/ or Kashin Beck disease/ or basilar impression/ or hyperostosis/ or enchondromatosis/	14468
48	(osteochondrodysplasia* or dyschondroplasia* or dyschondrosteos* or chondrodystroph* or spondylodysplast* or spondyloepiphyseal dysplasia* or (spondylo-epi* and dysplasia*) or spondyloepiphyseal dysplasia congenita or SEDC or SEMD or (Kniest adj (dysplasia or syndrome*)) or Melnick-Needles syndrome* or mucopolysaccharidos* or MPS IV or eccentroosteochondrodysplasia* or eccentro-osteochondrodysplasia* or (morquio* adj (disease or syndrome*)) or galns deficien* or galactosamine-6-sulfatase deficien* or hypochondrogenesis or fibrochondrogenesis or achondrogenesis or atelosteogenesis or diastrophic dysplasia or otospondylomegaepiphyseal dysplasia or dyssegmental dysplasia or Schneckenbecken or opsismodysplasia* or bent bone dysplasia* or Stuve-Wiedemann or ((conradi and hunermann) adj2 syndrome) or Greenberg dysplasia or acrodysplasia* or ((giedion and langer) adj syndrome) or tricho rhino phalangeal syndrome or trichorhinophalangeal syndrome or kashin beck disease or platybasia* or bone hypertroph* or (ollier* adj2 disease) or enchondrom* or chondroplasia angiomatosis or chondrodysplasia*).ti,kf.	14037
49	(otopalatodigital adj2 (spectrum or syndrome or disorder*)).ti,ab,kf.	75

50	(chondrodystroph* adj2 myoton*).ti,ab,kf.	43
51	(melnick adj2 needles adj (syndrome* or osteodysplasty)).ti,ab,kf.	112
52	(endosteal hyperostosis or hyperphosphatasemia tarda or ((schwartz-jampel* or sja) adj2 syndrome*) or x-linked sed* or sed tarda or multiple epiphyseal dysplasia* or hyperostosis or myotonic myopath* or van buchem disease or sclerosteosis or osteoporosis pseudoglioma or OPPG syndrome* or eosinophilic granuloma* or (engelmann* adj (disease or syndrome*))).ti,ab,kf.	15911
53	Ellis van Creveld syndrome/	887
54	(ellis van creveld adj2 (syndrome or dysplasia)).ti,ab,kf.	617
55	((chondroectodermal or mesoectodermal) adj2 dysplasia*).ti,ab,kf.	213
56	chondrodysplasia punctata/	1270
57	(chondrodysplasia or brachytelephalangic or ((conradi and hunermann) adj syndrome*)).ti,ab,kf.	2622
58	(CHILD syndrome or congenital hemidysplasia or fibula hemimelia* or (genu adj (valgum or varum or varus))).ti,ab,kf.	3038
59	slipped capital femoral epiphysis/	1838
60	((coxa adj (magna* or valga* or valgus or vara)) or slipped capital femoral epiphys*).ti,ab,kf.	3996
61	synostosis/	3227
62	(synostosis or synostoses or spondylocarp*).ti,ab,kf.	5298
63	acroosteolysis/	930
64	Hajdu Cheney syndrome/	281
65	(cheney syndrome or acroosteolysis or acro-osteolysis or serpentine fibula).ti,ab,kf.	1061
66	((hajdu and cheney) adj syndrome).ti,ab,kf.	231
67	pseudohypoparathyroidism/	2806
68	(pseudohypoparathyroidism* or pseudohypoparathyreoidism* or (albright adj2 osteodystroph*) or phd ib or phd1b or php ia or phpia).ti,ab,kf.	2677
69	(hypoparathyreoidism or hypoparathyroidism).ti,ab,kf.	11041
70	Paget bone disease/	9376
71	osteitis deformans.ti,ab,kf.	742
72	((paget* adj disease) and bone).ti,ab,kf.	5828
73	((methotrexate or MTX) adj osteopathy).ti,ab,kf.	44
74	ossifying myositis/	3863
75	osteolysis/	98611
76	(fibrodysplasia ossificans or myositis ossificans or ossifying myositis or ossifying fibrodysplasia or stone man syndrome* or osteoarthropathy or	8317

	paraosteoarthropathy or (Gorham-Stout adj (disease or syndrome)) or vanishing bone disease or melorheostos*).ti,ab,kf.	
77	fibrous dysplasia/	7908
78	((Albright* or (FD adj MAS)) adj3 (disease or syndrome* or spectrum or orosteodystrophia*)).ti,ab,kf.	2317
79	(fibrous dysplasia* or osteodysplasia* or osteodysplastic or fibrous osteodystrophy or osteodystrophia fibrosa or fibrodysplasia* or osteocraniostenos* or Desbuquois dysplasia*).ti,ab,kf.	9056
80	Albright syndrome/	5454
81	(osteitis fibrosa cystica or osteodystrophia fibrosa or bone fibrous dysplasia or dysplasia fibrosa or (Recklinghausen* adj disease)).ti,ab,kf.	4311
82	(fibrous bone adj (defect* or disease* or dysplasia*)).ti,ab,kf.	88
83	leg length inequality/	5574
84	(leg length inequalit* or femoral deficiency or congenital femoral deficien* or proximal focal femoral deficien*).ti,ab,kf.	676
85	(limb* adj3 (reduction adj2 (defect* or deform* or deficien*))).ti,ab,kf.	543
86	(arachnodactyl* or brachydactyl* or ectromelia* or amelia* or hemimelia* or phocomelia* or sirenomel* or (proteus adj2 syndrome)).ti,ab,kf.	7412
87	basal cell nevus syndrome/	3399
88	(basal cell nevus syndrome or basal cell carcinoma syndrome or nbccs or fifth phacomatos* or (gorlin adj2 syndrome)).ti,ab,kf.	2892
89	Camurati Engelmann disease/	396
90	(engelmann* adj (disease or syndrome)).ti,ab,kf.	451
91	(chondroectodermal dysplasia* or diaphyseal dysplasia* or diaphyseal hyperostos* or metaphyseal dysplasia* or Cartilage-hair hypoplasia*).ti,ab,kf.	1337
92	or/1-91	403205
93	child/ or infant/ or adolescent/ or pediatrics/ or "minor (person)"/ or (pediatric* or paediatric* or child* or infan* or toddler* or pubescen* or teen* or adolesc* or youth or (young adj (person* or patient*))).ti,ab,kf.	5513462
94	child health care/	41948
95	93 or 94	5517120
96	adult/ or (adult or adults or adulthood or maturity or mature or grown-up* or grown-up* or "coming of age" or lifespan or life span or lifetime or lifelong).ti,ab,kf.	12525788
97	disease management/ or personal autonomy/ or patient advocacy/ or patient participation/ or self care/ or self concept/ or decision making/ or shared decision making/ or health literacy/ or psychological adjustment/ or coping/	761104

98	((patient* or self* or personal) adj7 (autonom* or manag* or admin* or advoca* or engag* or involv* or participat* or activ* or proactiv* or cope or copes or coping or empower* or relian* or sufficien* or sustain* or litera* or efficac* or mastery or organiz* or organis* or adapt* or navigat* or agency or choice* or choos* or determin* or decis* or decid* or independ* or depend* or control* or passiv* or prefer* or govern* or follow-up or judg* or authorit* or unsupervis* or ((deal* or liv*) adj (with or through)))).ti,ab,kf.	4703483
99	or/96-98	15034575
100	95 and 99	2594842
101	disease exacerbation/	221015
102	((clinical or disease* or disorder* or illness* or condition*) adj2 (course* or progress* or process* or develop* or evolv* or exacerbat* or deterior* or trajector*)).ti,ab,kf.	961467
103	longitudinal study/	251175
104	(longitud* or long term or longterm).ti,kf.	550059
105	follow up/	2520949
106	((follow up or followup) adj (study or studies)).ti,ab,kf.	95302
107	socioeconomics/ or vulnerable population/ or poverty/ or social class/ or social support/ or financial management/ or health behavior/ or "social determinants of health"/ or health disparity/ or social problem/ or happiness/	714581
108	(socioeconomic* or SES or SEP or sociodemographic* or socio-demographic* or SDOH or income or wealth* or poverty or educational level or level of education or educational attainment or well educated or better educated or academic achievement* or unemploy* or home owner* or tenure or affluen* or well off or better off or worse off or ethnic* or race or racial* or racis* or equit* or inequit* or inequalit* or disparit* or equality).ti,ab,kf.	1399608
109	((social* or socio-economic or socioeconomic or economic or structural or cultural) adj3 (advantag* or disadvantag* or exclude* or exclusion or include* or inclusion or status or position* or gradient* or hierarch* or class* or determinant*)).ti,ab,kf.	244780
110	"quality of life"/	792265
111	("quality of life" or life quality or qol or hrqol or "hr-qol" or hrql or ((patient* or personal*) adj2 (satisf* or happiness))).ti,ab,kf.	905215
112	or/100-111	8289498
113	(continu* adj2 (care or healthcare)).ti,kf.	8251
114	transition to adult care/	3580
115	(transition care or healthcare transition or transition clinic* or "transfer of care").ti,ab,kf.	3671

116	((transit* or transfer*) adj3 (adult* or care or healthcare or clinic* or program* or care or process* or period or plan* or system* or strateg* or model* or phase or tool* or guideline* or recommendat* or standardi* or journey* or trajector*)).ti,kf.	40685
117	((transition* or transfer) adj2 (program* or care or healthcare or health-care or process* or period or plan* or strateg* or model* or system* or phase or tool* or guideline* or recommendat* or standardi* or journey* or trajector*)).ti,ab,kf.	125854
118	(transition* or transfer).ti.	282329
119	or/113-118	378865
120	92 and 112 and 119	608
124	limit 120 to (danish or english or french or german or norwegian or portuguese or spanish or swedish)	510

Scopus:

(((TITLE ((pediatric OR pediatrics OR paediatric OR paediatrics OR child* OR infant* OR infancy OR toddler* OR adolescent* OR adolescence OR pubescent* OR teenager* OR teen OR teens OR young person* OR youth) AND (adult OR adults OR adulthood OR maturity OR mature OR "grown up" OR "grown-up" OR "coming of age" OR lifespan OR "life span" OR lifelong OR lifetime))) OR (((TITLE-ABS-KEY ((longitudinal OR "long term" OR "long-term" OR longterm OR follow-up OR "follow up" OR followup) PRE/2 (study OR studies))) OR (TITLE-ABS-KEY ((clinical OR disease* OR disorder* OR illness* OR condition*) PRE/2 (course* OR progress* OR process* OR develop* OR evolv* OR exacerbat* OR deterior* OR trajector* OR amelior* OR alleviat* OR better* OR heal*))) OR (TITLE-ABS-KEY ((patient* OR self* OR personal) W/2 (autonom* OR manag* OR admin* OR advoca* OR engag* OR involv* OR participat* OR activ* OR proactiv* OR cope OR copes OR coping OR empower* OR litera* OR efficac* OR master* OR organiz* OR organis* OR adapt* OR navigat* OR agency OR choice* OR choos* OR determin* OR decis* OR decid* OR independ* OR depend* OR control* OR passiv* OR prefer* OR govern* OR judg* OR authorit* OR "deal with" OR "dealing with" OR "live with" OR "living with"))) OR (TITLE-ABS-KEY ({disease management} OR {autonomy} OR {autonomous} OR {adulthood} OR {adult care} OR {patient advocacy} OR {patient participation} OR {self concept} OR {self-concept} OR {self management} OR {self-management} OR {self-managing} OR {decision making} OR {decision-making} OR {choice behavior} OR {choice behaviour} OR {health literacy} OR {psychological adjustment} OR {coping} OR {cope} OR {copes} OR {empower} OR {empowers} OR {empowerment} OR {empowering} OR {empowered} OR {independent} OR {independence} OR {unsupervised}))) AND ((TITLE-ABS-KEY (pediatric OR pediatrics OR paediatric OR paediatrics OR child* OR infant* OR infancy OR toddler* OR adolescent* OR adolescence OR pubescent* OR teenager* OR teen OR teens OR young person* OR youth)) AND (TITLE-ABS-KEY (adult OR adults OR adulthood OR maturity OR mature OR "grown up" OR "grown-up" OR "coming of age" OR lifespan OR "life span" OR lifelong OR

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Table S2: Summary of the 34 articles included in the scoping review

Main author last name (year)	Country(s)	Name of the RBMC(s)	Study design	Study aim	Study sample	Main findings
Ricks (2025)	United States	Achondroplasia, OI, SEMD, SEDT, Acromicric/ geleophysic dysplasia	Qualitative study (semi-structured interview)	To understand and describe how young adults with short stature and skeletal dysplasia define independence, the barriers they face in achieving and maintaining independence, and the resources they leverage to support their independence during the transition to adulthood	Patients (10)	During the transition to adulthood, the independence of young adults with skeletal dysplasia is shaped by the conflict between the desire for autonomy and physical limitations. Supportive environments, adaptive tools, and peer networks are essential. Providers play a key role in facilitating independence through guidance, accessibility, and connection to resources
Grasemann (2025)	Germany, Netherlands, Denmark, UK, Austria, Turkey, Norway, Portugal, France, Greece, UK	RBMCs (in general)	Expert opinion/ consensus	To develop consensus-based recommendations for transition care in RBMCs, focusing on best practices for seamless transition and patient empowerment	HCPs; Patient advocacy groups	Structured, patient-centered, multidisciplinary transition care is essential for RBMCs. Empowerment, coordination, and early preparation are key. Future research should evaluate feasibility and real-world impact
Celli (2024)	Netherlands, Italy, Norway, Belgium, Germany, Estonia	OI	Systematic review	To summarize the literature on OI patient pediatric to adult care transition experiences and compare OI transition approaches to other chronic disease	NA	Transition in OI remains poorly defined and inconsistently implemented. Requires structured, multidisciplinary, personalized approaches with strong communication and self-management support. More research is needed to evaluate gradual transition models adapted to OI. Five main steps were identified for a structured and adapted OI transition: Education; Readiness; Preparation; Transition; and Follow-up

(continues)

Table S2: Summary of the 34 articles included in the scoping review

Main author last name (year)	Country(s)	Name of the RBMC(s)	Study design	Study aim	Study sample	Main findings
Stepien (2024)	Netherlands, UK, United States	Gaucher disease type 1	Qualitative study (survey)	To evaluate the transition process in various countries and to understand the challenges that both healthcare professionals and patients experience	Patients (45); HCPs	Transition processes for GD1 are inconsistent and underdeveloped globally. Only 33% of patients were aware of transition clinics. Healthcare professionals also reported limited availability of transition clinics, inconsistent protocols, and challenges such as poor coordination, lack of expertise in adult services, and insufficient funding. A standardized, well-coordinated, multidisciplinary approach is needed to support adolescents moving into adult care
Scognamiglio (2024)	Italy	MO, Ollier disease, Maffucci syndrome	Qualitative study: focus groups	To investigate the priorities or patients, families, caregivers, and health care professionals regarding the transition process from child to adult care in MO, Ollier disease, and Maffucci syndrome	Patients (30); Parents (61); HCPs	The study highlights the central role of stakeholder engagement in shaping transition processes and informing future protocol development. Effective transition depends on patient self-advocacy supported by core elements such as a structured care plan, general practitioner involvement, and digital health records. Ultimately, coordinated input from patients, families, providers, and psychological services is essential
Kastelic (2024)	Latin-American countries	XLH	Expert opinion/ consensus	To provide a set of recommendations to facilitate the transition process and ensure adequate management and follow-up, as well as the quality of life for patients with XLH as they transition to adult life	HCPs	Transition in XLH is complex and requires structured, multidisciplinary, culturally adapted approaches. Psychosocial and medical readiness must guide timing. Strong emphasis on patient autonomy, education, and continuity of care

(continues)

Table S2: Summary of the 34 articles included in the scoping review

Main author last name (year)	Country(s)	Name of the RBMC(s)	Study design	Study aim	Study sample	Main findings
Fredwall (2024)	Norway, Saudi Arabia, Portugal, Qatar, Italy, France, Germany, Spain, UK	Achondroplasia	Expert opinion/ consensus	To develop a patient-held checklist to support adults with achondroplasia to be delivered to young people with achondroplasia during the transition from pediatric to adult care	HCPs	Transition should begin early, with education and empowerment of the person with achondroplasia. Overall, the paper advocates for a proactive, structured, and patient-centered approach to adult achondroplasia care, with the checklist acting as a bridge between pediatric expertise and adult healthcare realities
Munns (2023)	Australia, China, Hong Kong, Japan, Malaysia, Singapore, South Korea, Taiwan, Thailand	XLH	Expert opinion/ consensus	To use the Delphi methodology to develop the most up-to-date XLH consensus recommendations on the diagnosis, multidisciplinary management, and transition from pediatric to adult care, specifically tailored to the Asia-Pacific Countries setting	HCPs	A structured transition process from pediatric to adult services is essential, beginning in adolescence and supported by readiness assessments, clear communication between teams, and continuity of monitoring for pain, mobility, and dental health. The article also calls for improved clinician education across Asia-Pacific region to reduce diagnostic delays and enhance long-term outcomes
Ross (2023)	United states	XLH, OI, Hypophosphatasia	Literature review	To provide a brief review of research and guidelines for transitions of care more generally, followed by a more detailed treatment of bone disorders specifically	NA	Existing gaps in guidelines and limited evidence highlight the need for ongoing research and strategic development of transition pathways to enhance patient outcomes and quality of life. Robust infrastructure and multidisciplinary teams are needed to ensure effective transition for patients with metabolic bone disorders. And expanding telehealth services could help promote equitable access

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Main author last name (year)	Country(s)	Name of the RBMC(s)	Study design	Study aim	Study sample	Main findings
Rauch (2023)	Canada	OI	Literature review	To review issues regarding the transition from pediatric to adult-focused health care for individuals with OI	NA	Although self-management resources are available, variation in transition practices across healthcare systems limits their broader applicability. As individuals with OI age, their clinical presentation changes, with fewer fractures but a greater burden of extraskeletal complications. Developing clearer guidelines for adult services would strengthen the pediatric-to-adult transition and better accommodate these evolving needs
Casado (2023)	Spain	XLH	Expert opinion/consensus: task force discussions	To assess HCPs' experiences with transitioning patients with metabolic bone diseases in Spain and to develop consensus recommendations with the specialists involved in their care	HCPs	The transition of care for people with XLH in Spain is inconsistent and underdeveloped. Clear need for structured, multidisciplinary, well-coordinated transition programs
Zuber (2023)	Poland	Muco-polysaccharidosis	Literature review	To describe methods of diagnosis, treatment, and management of patients with Mucopolysaccharidosis	NA	The authors emphasize the need for structured transition pathways, improved communication between pediatric and adult providers, and the development of adult rare disease centers. Patient associations play a vital role in supporting families and facilitating access to expertise
Fredwall (2022)	Norway, Saudi Arabia, Portugal, Qatar, Italy, France, Germany, Spain, UK	Achondroplasia	Qualitative study (survey)	To understand current practices in the transition process of individuals with achondroplasia from pediatric to adult services and how adults perceive their care	HCPs; Patient advocacy groups	Half of those surveyed rated the transition experience to adult services as inadequate. Transition is poorly coordinated and contributes to disengagement. Lifelong, proactive, multidisciplinary care is essential but not yet widely implemented

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Main author last name (year)	Country(s)	Name of the RBMC(s)	Study design	Study aim	Study sample	Main findings
Kubota (2022)	Japan	XLH	Literature review	To describe transitional care and team management for patients with XLH	NA	XLH requires lifelong, coordinated, multidisciplinary care, and structured transition is essential to maintain health, prevent complications, and support autonomy in adulthood
Hill (2022)	UK	OI	Qualitative study (semi-structured interview)	To improve understanding of the impact of OI on the daily lives of individuals and families and to consider how the condition is managed so that support needs can be better addressed	Patients (17); Parents (8); HCPs; Patient advocacy groups	The study identifies significant gaps in adult care, particularly in coordination and psychosocial support, compared to pediatric services. Professionals highlight the need for improved mental health support and better transition planning
Dahir (2022)	United States	XLH	Expert opinion/consensus	To review the literature and to provide the expert consensus recommendations on health care transition preparation for adolescents and young adults with XLH from pediatric- to adult-focused health care developed by clinical care investigators and transition experts	HCPs	XLH requires a tailored, structured transitional care program to prevent loss to follow-up and ensure lifelong care. Clear guidelines, individualized planning, and multidisciplinary collaboration are essential. Effective transition requires early preparation, disease-specific education and strong communication between pediatric and adult teams
Cormier-Daire (2021)	France, Saudi Arabia, Qatar, Portugal, Italy, Norway, Germany, UK, Belgium	Achondroplasia	Expert opinion/consensus	To develop a consensus on guiding principles of management of achondroplasia to provide a basis for developing optimal care in Europe	HCPs	Transition is explicitly recognized as a critical component of lifelong care. Requires coordination, continuity, and attention to psychosocial well-being. Adult services must be prepared to manage complex, evolving needs

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Main author last name (year)	Country(s)	Name of the RBMC(s)	Study design	Study aim	Study sample	Main findings
Amajjar (2021)	Netherlands	MO	Qualitative study (semi-structured interview)	To gain insight on how to organize the transition from pediatric to adult care for patients with MO	Patients (12); Parents (10); HCPs	The transfer process for MO patients is currently unsatisfactory. Improvements require: early preparation, self-management support, psychological care, and coordinated multidisciplinary collaboration
Shaw (2021)	UK	OI	Qualitative study: focus groups	To provide a detailed insight into how parents understand and experience their caregiving role during their child's transition to adult services, to identify parents needs, and to inform service improvements	Parents (13)	A more holistic framework that accounts for how families interpret vulnerability and risk would strengthen transitional care. Participants identified the need for clearer guidance and more coordinated services to manage risk and uncertainty during routine transitions. Parents' reliance on protective practices, though well-intentioned, can inadvertently restrict adolescents' autonomy as they enter midlife. Achieving an effective balance between immediate health protection and long-term wellbeing remains essential, and integrating collaborative, proactive planning with families should be considered a core component of transitional care
Giannini (2021)	Italy	XLH	Systematic review	To review the available clinical evidence and discuss the current challenges underlying the transition from childhood to adulthood care to develop appropriate management and follow-up patterns in adult XLH patients	NA	The review highlights major gaps in transition and adult care. Many young adults discontinue treatment after leaving pediatric services, often due to poor tolerability of conventional therapy, lack of specialist follow-up, or inconsistent clinical advice. Structured transition, multidisciplinary adult care, and clearer therapeutic guidance are urgently needed to reduce long-term morbidity

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Main author last name (year)	Country(s)	Name of the RBMC(s)	Study design	Study aim	Study sample	Main findings
Huurneman (2021)	Netherlands	tURLD	Qualitative study: focus groups	To evaluate the role of the rehabilitation care in assisting young adults with a tULRD and their parents in dealing with limitations during the transition from adolescence to adulthood	Patients (16); Parents (11); HCPs	The findings align with self-determination theory, emphasizing the importance of competence, relatedness, and autonomy during the transition to adulthood
Schnabel (2020)	Germany	XLH	Expert opinion/consensus	To show the need of a biopsychosocial concept of care in the transition process and later	HCPs	XLH requires a structured, biopsychosocial, multidisciplinary transition care model. Long-term care and transition planning are essential for maintaining participation and quality of life. The article highlights the importance of a planned transition to adult care to ensure continuity of management for this lifelong condition
Michalovic (2020)	Canada	OI	Qualitative study (semi-structured interview)	To explore the perceived self-management needs of young adults with OI with the goal of optimizing the self-management and transitional care services	Patients (7)	Improving self-management and transitional care for young people with OI requires stronger collaboration among all stakeholders. This need is underscored by adolescents' reports of frustration with the transition process. Addressing these challenges depends on adequately preparing adult-care clinicians and providing adolescents and young adults with targeted education and mentorship

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Main author last name (year)	Country(s)	Name of the RBMC(s)	Study design	Study aim	Study sample	Main findings
Lampe (2019)	Germany, Spain, Russia, UK	Muco-polysaccharidosis	Case studies	To describe case studies from four European inherited metabolic disease centers to showcase examples of transition strategies used for patients with Mucopolysaccharidosis	Patients (7)	Across four European centers, transition strategies varied widely, but several elements consistently supported success: starting early, providing clear explanations, offering psychological support, and ensuring repeated contact with adult clinicians. Overall, transition must be individualized, flexible, and tailored to each patient's medical, emotional, and social needs. Strong multidisciplinary team coordination, and patient-centered communication are essential for success
Koumakis (2019)	France	OI, Achondroplasia, MO	Expert opinion/consensus	To summarize the knowledge about transitional care in rare skeletal dysplasia	HCPs	Transition must be gradual, personalized, and coordinated. Joint consultations between pediatric and adult teams are key. New tools and structures are needed to prevent loss to follow-up
Haffner (2019)	Germany, France, Italy, Sweden, UK, Belgium	XLH	Expert opinion/consensus: guidelines	To develop clinical practice recommendations for the diagnosis and management of patients with XLH	HCPs; Patient advocacy groups	The guideline stresses the importance of structured transition to adult care and that continuity of monitoring and treatment is essential. Genetic counselling is recommended at transition, adult care must address chronic pain, mobility limitations, pseudofractures, enthesopathies, and dental issues

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Main author last name (year)	Country(s)	Name of the RBMC(s)	Study design	Study aim	Study sample	Main findings
Jeong (2019)	Canada	OI	Expert opinion/consensus: task force discussions	To develop an evidence-based passport for individuals with OI. In particular, to systematically review and synthesize studies about: a) the Good2Go MyHealth Passport Program; b) use of any portable, health related tools for care transitions and; c) the care transitions for individuals with OI	Patients (1); HCPs	The 'OI MyHealth Passport' facilitates the transition of care for individuals with OI by supporting self-advocacy and continuity of care
Carrier (2018)	Canada	OI	Expert opinion/consensus: expert task force	To synthesize existing literature, establish evidence-based transition guidelines, and create a transfer tool tailored to adolescents with OI	Patients (2); HCPs; Social workers; Administrative staff	Designed to improve adolescents' transition experiences, the 'OI Transfer Summary' functions as a person-centered resource that facilitates a smoother transition from pediatric to adult services. By providing structured guidance, it supports a more coordinated and seamless transfer of care
Ghotme (2018)	Argentina, Brazil, Canada, Colombia, Ireland, Spain, United Arab Emirates, United States	Muco-polysaccharidosis	Qualitative study (survey)	To assess the challenges of managing spinal cord issues in adult patients with Mucopolysaccharidosis and how they affect the transition of care	HCPs	Effective transition requires structured planning, knowledge transfer, patient and family education, and institutional support. Overall, the findings call for the development of clear, coordinated guidelines to ensure safe and consistent care for adults with Mucopolysaccharidosis

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Main author last name (year)	Country(s)	Name of the RBMC(s)	Study design	Study aim	Study sample	Main findings
Couce (2018)	Spain	Muco-polysaccharidosis	Expert opinion/consensus	To create a number of recommendations for pediatricians, internists and other health professionals regarding the transition from pediatric to adult care for patients with Mucopolysaccharidosis	HCPs	Begin preparation in early adolescence, ensure flexibility, individualize the plan, designate an adult care coordinator and maintain continuity, and establish a standardized pediatric discharge report. Ultimately, the goal is to maximize patient autonomy and ensure uninterrupted, developmentally appropriate care as individuals with MPS enter adulthood
Lankhorst (2017)	Netherlands	tULRD	Qualitative study (semi-structured interview)	To determine whether young adults with tULRD have experienced limitations in various domains of participation during transition to adulthood and how they dealt with these limitations	Patients (15)	The major challenge is vocational transition, where external judgments and limited opportunities create barriers. Rehabilitation teams could provide more structured support during this phase.
Hoyer-Kuhn (2017)	Germany	OI, XLH	Expert opinion/consensus: task force discussions	Show the need of continuous care when there is no cure for the diagnosis	HCPs	Transition planning should be early, structured, and individualized, including joint consultations between pediatricians and adults and the development of centralized adult care centers for rare skeletal diseases

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Main author last name (year)	Country(s)	Name of the RBMC(s)	Study design	Study aim	Study sample	Main findings
Dogba (2014)	Canada	OI	Qualitative study (semi-structured interview)	To evaluate a transition program for adolescents and young adults with OI from a pediatric orthopedic hospital to adult care with the aim of making recommendations for its improvement	Patients (6); Parents (4); HCPs; Social workers; Administrative staff	The strengths identified include a strong theoretical foundation built on parent–provider partnership and a comprehensive framework that promotes independent living and vocational integration. Identified weaknesses involve the discontinuation of care, organizational restructuring, and potential tensions between the transition program and participation in research protocols. A transition model for individuals with OI must be tailored, theoretically grounded, and responsive to patient preferences while remaining aligned with organizational practices and subject to ongoing evaluation
Shapiro (2012)	United States	OI	Expert opinion/consensus	To provide guidelines for pediatricians, adult physicians in different medical disciplines and patients families who are planning the transition to adult care for the adolescent with OI	HCPs	Four key topics related to effecting the transition from pediatric to adult care were discussed in this article: 1) Transitioning and maintaining health, 2) Preserving or improving the level of function, 3) Assuring continuity of medical/surgical care, and 4) Re-structuring psychosocial and work-related systems that previously were family-centric. Effective transition depends on strong communication between pediatric and adult healthcare teams, along with proactive engagement from the patient and their family. Patients are supported in taking an active role as advocates and coordinators of their own care

RBMCs: rare bone mineral conditions. HCPs: healthcare professionals. GD1: Gaucher disease type 1. XLH: X-linked Hypophosphatemia. OI: Osteogenesis imperfecta. MPS: Mucopolysaccharidosis. tULRD: Transversal upper limb reduction deficiency. MO: Multiple osteochondromas. SEMD: spondyloepimetaphyseal dysplasia. SEDT: spondyloepiphyseal dysplasia tarda.