Annexe 1: Articles source des case reports avec les signes et symptômes introduits dans SYNDROC pour chaque case report.

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
Acrorenoocular	A	Aalfs CM, van Schooneveld MJ, van Koulen EM, Hennekam RC. Further delineation of the acro-renal-ocular syndrome. Am J Med Genet. Wiley Subscription Services, Inc., A Wiley Company; 1996 Mar 29;62(3):276–81.	Case: "...Temtamy et al 1...."	...Ear malformation...	Ear anomaly
				...Contracture...	Joint contracture
				...Radius hypoplasia/aplasia...	Radius hypoplasia
				...Renal malrotation...	Renal anomaly
				...Thumb hypoplasia/aplasia...	Thumb hypoplasia
	B		Case: "...Naito et al 1...."	...Coloboma...	Colobomata of iris
				...Nystagmus...	Nystagmus
				...Preaxial polydactyly...	Preaxial polydactyly
				...Renal malrotation...	Renal anomaly
				...Ventricular septal defect...	Ventricular septum defect
Alagille	A	Izumi K, Hayashi D, Grochowski CM, Kubota N, Nishi E, Arakawa M, et al. Discordant clinical phenotype in monozygotic twins with Alagille syndrome: Possible influence of non-genetic factors. Am J Med Genet. 2015 Oct 13;170(2):471–5.	Case: "...Twin A..."	...jaundice...	Jaundice
				...peripheral pulmonic steno- sis...	Peripheral pulmonary arterial stenosis
				...upslanted palpe- bral fissures...	Upward slanted palpebral fissure
				...A butterfly vertebra...	Vertebral anomaly
	B	Li PH, Shu SG, Yang CH, Lo FC, Wen MC, Chi CS. Alagille syndrome with interstitial 20p deletion derived from maternal ins(7;20). American Journal of Medical Genetics. Wiley Subscription Services, Inc., A Wiley Company; 1996 Jun 28;63(4): 537–41.	Case: "...The propositus..."	"...intracellular and intracanalicular cholestasis..."	Cholestasis
				"...delayed bone age (only at 3 years of age)..."	Delayed osseous maturation
				"...anti-mon- goloid slant of palpebral fissures..."	Downward slanted palpebral fissure
				"...frontal bossing..."	Frontal bossing
				"...The liver was firm and en larged,..."	Hepatomegaly
				"...1), mild hypertelorism..."	Hypertelorism
				"...persistent jaundice..."	Jaundice
				"...head cir cumference of 47 cm (-3.1 SD)..."	Microcephaly
				"...markedly short and thin child..."	Short stature
				"...soft tissue swelling..."	Soft tissue swelling
Alstrom	A	Awazu M, Tanaka T, Sato S, Anzo M, Higuchi M, Yamazaki K, et al. Hepatic dysfunction in two sibs with Alström syndrome: case report and review of the literature. Am J Med Genet. 1997 Mar 3;69(1):13–6.	Case: "...Horiuchi et al. [1976], F..."	Table 1: "...Nerve deafness..."	Deafness
				Table 1: "...NIDDM..."	Diabetes mellitus
				Table 1: "...Obesity (degree)..."	Obesity
				Table 1: "...Retinal degeneration..."	Retinal degeneration
	B		Case: "...Patient 2, F..."	Table 1: "...Acanthosis nigricans..."	Acanthosis nigricans
				Table 1: "...Nerve deafness..."	Deafness
				Table 1: "...NIDDM..."	Diabetes mellitus
				Table 1: "...Obesity (degree)..."	Obesity
				Table 1: "...Retinal degeneration..."	Retinal degeneration
Arthrodentoosteo dysplasia	A	Brennan AM, Pauli RM. Hajdu--Cheney syndrome: evolution of phenotype and clinical problems. American Journal of Medical Genetics. 2001 May 15;100(4):292–310.	Case: "...Case 1..."	"...Fifth finger clinodactyly was present in both hands...."	Clinodactyly
				"...downslanting of palpebral fissures..."	Downward slanted palpebral fissure
				"...hypoplasia of the tooth enamel..."	Enamel hypoplasia
				"...bilateral epicanthic folds..."	Epicanthus
				"...slightly large head with frontal bossing...."	Frontal bossing
				"...Generalized hirsutism was present..."	Hirsutism
				"...metacarpal-phalangeal hypermobility (at age 4 years),..."	Joint hypermobility
				"...height had dropped to the 5th centile and she remains mildly short statured at age 5 years, with a height of 103.5cm (5th centile)...."	Short stature
				"...mild irregularity of the brows with synophrys...."	Synophrys
				"...multiple wormian bones..."	Wormian bone
	B		Case: "...Case 3..."	"...submucous cleft palate..."	Cleft palate
				"...diastasis of the sagittal suture...."	Delayed closure of fontanelle
				"...downslanting palpebral fissures..."	Downward slanted palpebral fissure
				"...frontal bossing..."	Frontal bossing
				"...hypoplastic frontal sinuses..."	Frontal sinus hypoplasia
				"...right multicystic dysplastic kidney..."	Kidney dysplasia
				"...and microretrognathia..."	Micrognathia
				"...patent ductus arteriosus necessitating ligation..."	Patent ductus arteriosus
				"...synophrys..."	Synophrys
				"...wormian bones..."	Wormian bone
Asphyxiating thoracic dysplasia	A	Keppler-Noreuil KM, Adam MP, Welch J, Muilenburg A, Willing MC. Clinical insights gained from eight new cases and review of reported cases with Jeune syndrome (asphyxiating thoracic dystrophy). Am J Med Genet. 2011 Apr 4;155(5):1021–32.	Case: "...Case 1..."	Table 1: "...Brachydactyly/micromelia..."	Brachydactyly
				Table 1: "...Limb shortening/rhizomelia..."	Rhizomelic limb
				Table 1: "...Limb shortening/rhizomelia..."	Short limb
				Table 1: "...Short stature (< 3rd centile)..."	Short stature
				Table 1: "...Narrow/small chest (CC)..."	Small thorax
	B		Case: "...Case 2..."	Table 1: "...Brachydactyly/micromelia..."	Brachydactyly
				Table 1: "...Cholestasis/liver dysfunction (resolved by 21 months)..."	Cholestasis
				Table 1: "...Limb shortening/rhizomelia..."	Rhizomelic limb
				Table 1: "...Limb shortening/rhizomelia..."	Short limb
				Table 1: "...Narrow/small chest (CC)..."	Small thorax
Barber-say	A	Dinulos MB, Pagon RA. Autosomal dominant inheritance of Barber-Say syndrome. Am J Med Genet. 1999 Sep 3;86(1):54–6.	Case: "...Sod et al. [1997]..."	Table 1: "...Small, dysplastic ears..."	Ear anomaly
				Table 1: "...Ocular hypertelorism..."	Hypertelorism
				Table 1: "...Macrostomia..."	Macrostomia
				Table 1: "...Developmental delay/MR..."	Mental deficiency
				Table 1: "...Redundant skin..."	Redundand skin
	B		Case: "...Patient 2..."	Table 1: "...Hearing loss..."	Deafness
				Table 1: "...Small, dysplastic ears..."	Ear anomaly
				Table 1: "...Ocular hypertelorism..."	Hypertelorism
				Table 1: "...Macrostomia..."	Macrostomia
				Table 1: "...Redundant skin..."	Redundand skin
				Table 1: "...Shawl scrotum..."	Shawl scrotum
Brachmann-de lange	A	Manouvrier S, Espinasse M, Vaast P, Boute O, Farre I, Dupont F, et al. Brachmann-de Lange syndrome: pre- and postnatal findings. Am J Med Genet. Wiley Subscription Services, Inc., A Wiley Company; 1996 Mar 29;62(3):268–73.	Case: "...Case 3..."	"...long bulging philtrum..."	Long philtrum
				"...with severely depressed nasal bridge..."	Low nasal bridge
				"...severe micrognathia..."	Micrognathia
				"...IUGR (biparietal diam eter, abdominal circumference, and long bone measure ments <10th centile)..."	Prenatal onset growth deficiency
				"...radial hypoplasia..."	Radius hypoplasia
				"...presence of 13 ribs..."	Rib anomaly
				"...small nose..."	Small nose
				"...a small ventricular septal de fect..."	Ventricular septum defect
	B		Case: "...Case 2..."	"...fetus also presented with bilateral cryptorchidism..."	Cryptorchidism
				"...hypertrichosis..."	Hirsutism
				"...long eye lashes,..."	Long eyelash
				"...fetus with IUGR..."	Prenatal onset growth deficiency
				"...hypoplastic left forearm..."	Short forearm
				"...small nose..."	Small nose
				"...synophris..."	Synophrys
Cantu	A	Scurr I, Wilson L, Lees M, Robertson S, Kirk E, Turner A, et al. Cantú syndrome: report of nine new cases and expansion of the clinical phenotype. Am J Med Genet. 2011 Mar;155A(3):508–18.	Case: "...Patient 7..."	Table 1: "...Broad ribs..."	Broad rib
				Table 1: "...Coarctation..."	Coarctation of aorta
				Table 1: "..."Coarse" facial appearance..."	Coarse facies
				Table 1: "...Mild LV dilatation..."	Congenital heart defect
				Table 1: "...Hepatomegaly (1st few years)..."	Hepatomegaly
				Table 1: "...Hypertelorism..."	Hypertelorism
				Table 1: "...Recurrent infections..."	Immunity deficiency
				Table 1: "...Head circumference > 97th..."	Macrocephaly
				Table 1: "...PDA..."	Patent ductus arteriosus
				Table 1: "...Thick calvaria..."	Thick calvarium
	B		Case: "...Patient 9..."	Table 1: "...Wide metaphyses..."	Broad metaphysis
				Table 1: "...Broad ribs..."	Broad rib
				Table 1: "..."Coarse" facial appearance..."	Coarse facies
				Table 1: "...Hypertelorism..."	Hypertelorism
				Table 1: "...Long philtrum..."	Long philtrum
				Table 1: "...Flat nasal bridge (infancy)..."	Low nasal bridge
				Table 1: "...Mild motor and speech delay..."	Mental deficiency
				Table 1: "...Height < 3th..."	Short stature
				Table 1: "...Hyper-pigmentation..."	Skin hyperpigmentation
				Table 1: "...Chronic enlargement of salivary glands and elevated ESR and CRP, umbilical hernia..."	Umbilical hernia
Cardio-facio-cutaneous	A	Wong Ramsey KN, Loichinger MH, Slavin TP, Kuo S, Seaver LH. The perinatal presentation of cardiofaciocutaneous syndrome. Am J Med Genet. 2014 Apr 9;164(8):2036–42.	Case: "...Patient 2..."	"...hypertrophic cardiomyopathy involving the inter-ventricular septum with lesser involvement of the posterior wall of the left ventricle...."	Congenital heart defect
				"...non-descended right testis..."	Cryptorchidism
				"...posteriorly angulated ears..."	Ear anomaly
				"...mild hepatomegaly..."	Hepatomegaly
				"...appar- ent hypertelorism..."	Hypertelorism
				"...hypotonia..."	Hypotony
				"...severe global develop- mental delay..."	Mental deficiency
				"...polyhydramnios..."	Polyhydramnios
				"...redundant nuchal skin extending to upper shoulders,..."	Redundand skin
				"...failure to thrive..."	Short stature
	B	Karaer K, Lissewski C, Zenker M. Familial cardiofaciocutaneous syndrome in a father and a son with a novel MEK2 mutation. Am J Med Genet. 2014 Dec 8;167(2):385–8.	Case: "...A 4-month-old boy..."	...Atrial septal defect...	Atrial septum defect
				...cerebellar and frontotemporal atrophy...	Cerebral atrophy
				...CHDs...	Coronary disease
				...low-set and posteriorly rotated ears...	Ear anomaly
				...frontal bossing...	Frontal bossing
				...supraorbital hypoplasia...	Hypoplastic supraorbital ridge
				...low-set and posteriorly rotated ears...	Low-set ear
				...mild to moderate delay in psychomo- tor development....	Mental deficiency
				...micrognathia...	Micrognathia
				...polyhydramnios...	Polyhydramnios
Cerebral Gigantism	A	Balasubramanian M, Shearing E, Smith K, Chavasse R, Taylor R, Tatton-Brown K, et al. Pneumothorax from subpleural blebs-A new association of sotos syndrome? Am J Med Genet. 2014 Jan 23;164(5):1222–6.	Case: "...Patient 1..."	"...hypertelorism..."	Hypertelorism
				"...recurrent episodes of cough and fever..."	Immunity deficiency
				"...left inguinal hernia..."	Inguinal hernia
				"...developed polycythemia and jaundice..."	Jaundice
				"...flat nasal bridge..."	Low nasal bridge
				"...macrocephaly..."	Macrocephaly
				"...months, rolled over at 21 months and had very few discernible noises to communicate his needs. At 3 years, he was trying to pull to standing, babbled and had some head control...."	Mental deficiency
				"...polyhydramnios..."	Polyhydramnios
				"...umbilical hernia..."	Umbilical hernia
	B	Corsello G, Giuffrè M, Carcione A, Cuzto ML, Piccione M, Zino O. Lymphoproliferative disorders in Sotos syndrome: observation of two cases. Am J Med Genet. Wiley Subscription Services, Inc., A Wiley Company; 1996 Sep 6;64(4):589–93.	Case: "... Additionally observed patients: 3 of 3..."	Table 1: "...Cardiac murmur..."	Congenital heart defect
				Table 1: "...Abnormalities of external ears..."	Ear anomaly
				Table 1: "...Frontal bossing..."	Frontal bossing
				Table 1: "...Macrocephaly..."	Macrocephaly
				Table 1: "...Redundant skin..."	Redundand skin
				Table 1: "...Shawl scrotum..."	Shawl scrotum

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
				Table 1: "...Psychomotor delay..."	Mental deficiency
				Table 1: "...Prognathism..."	Prognathism
Coffin-siris	A	Fleck BJ, Pandya A, Vanner L, Kerkering K, Bodurtha J. Coffin-Siris syndrome: Review and presentation of new cases from a questionnaire study. American Journal of Medical Genetics. John Wiley & Sons, Inc; 2001 Feb 15;99(1):1–7.	Case: "...Patient 11..."	Table 1: "...Coarse appearance..."	Coarse facies
				Table 1: "...Cardiac findings..."	Congenital heart defect
				Table 1: "...Delayed bone age..."	Delayed osseous maturation
				Table 1: "...Abnormal/delayed dentition..."	Dental anomaly
				Table 1: "...Absent/hypoplastic fifth phalanx (hands)..."	Distal phalangeal hypoplasia
				Table 1: "...Hirsutism..."	Hirsutism
				Table 1: "...Frequent infections..."	Immunity deficiency
				Table 1: "...Macroglossia..."	Macroglossia
				Table 1: "...Absent/hypoplastic fifth finger /toenails..."	Nail hypoplasia
				Table 1: "...Short stature..."	Short stature
	B		Case: "...Patient 5..."	Table 1: "...Cleft palate..."	Cleft palate
				Table 1: "...Cardiac findings..."	Congenital heart defect
				Table 1: "...Hearing loss..."	Deafness
				Table 1: "...Abnormal/delayed dentition..."	Dental anomaly
				Table 1: "...Absent/hypoplastic fifth phalanx (hands)..."	Distal phalangeal hypoplasia
				Table 1: "...Abnormal ears..."	Ear anomaly
				Table 1: "...Hirsutism..."	Hirsutism
				Table 1: "...Frequent infections..."	Immunity deficiency
				Table 1: "...Absent/hypoplastic fifth finger /toenails..."	Nail hypoplasia
				Table 1: "...Renal findings..."	Renal anomaly
Costello	A	Gripp KW, Sol-Church K, Smpokou P, Graham GE, Stevenson DA, Hanson H, et al. An attenuated phenotype of Costello syndrome in three unrelated individuals with a HRASc.179G>A (p.Gly60Asp) mutation correlates with uncommon functional consequences. Am J Med Genet. 2015 Apr 25;167(9):2085–97.	Case: "...Individual 2 (CS#442)..."	..."blue episodes" ultimately attributed to a transient cardiac arrhythmia...	Arrythmia
				...tall forehead...	Broad forehead
				...attention deficit hyperactivity disorder and oppositional defiant disorder...	Hyperactivity
				...hypotonia was noted...	Hypotony
				...Significant hyperextensibility of the small joints of the hands were noted....	Joint hypermobility
				...OFC 55.8 cm (just above 98th centile)...	Macrocephaly
				...delays in fine motor skills, language acquisition, and social skills were identified....	Mental deficiency
				...collection of small tag-like skin papules involving the right areola and chest medial to the right nipple...	Papular skin lesion
	B	Mori M, Yamagata T, Mori Y, Nokubi M, Saito K, Fukushima Y, et al. Elastic fiber degeneration in Costello syndrome. Am J Med Genet. Wiley Subscription Services, Inc., A Wiley Company; 1996 Feb 2;61(4):304–9.	Case: "...Patient 1..."	"...bifid uvula..."	Bifid uvula
				"...various clinical findings such as "coarse"face (Fig. la) with prominent forehead..."	Coarse facies
				Table 1: "...Downslanting palpebral fissures..."	Downward slanted palpebral fissure
				"...epicanthal folds..."	Epicanthus
				"...hy- pertelorism..."	Hypertelorism
				"...depressed nasal bridge..."	Low nasal bridge
				Table 1: "...Macrocephaly..."	Macrocephaly
				Table 1: "...Macroglossia..."	Macroglossia
				Table 1: "...Large mouth..."	Macrostomia
				"...height was 61.0 cm (-2.9 SD)..."	Short stature
Cranio-cerebello-cardiac	A	Kosaki K, Curry CJ, Roeder E, Jones KL. Ritscher-Schinzel (3C) syndrome: documentation of the phenotype. Am J Med Genet. 1997 Feb 11;68(4):421–7.	Case: "...10 Digilio et al. [1995]..."	Table 1: "...Prominent forehead..."	Broad forehead
				Table 1: "...CAVC..."	Congenital heart defect
				Table 1: "..."Dandy-Walker malformation/variant"..."	Dandy-walker malformation
				Table 1: "...Downslanting palpebral fissures..."	Downward slanted palpebral fissure
				Table 1: "...Tetralogy of Fallot..."	Fallot
				Table 1: "...Hydrocephalus/enlarged lateral ventricles..."	Hydrocephalus
				Table 1: "...Ocular hypertelorism..."	Hypertelorism
				Table 1: "...Hypotonia..."	Hypotony
				Table 1: "...Macrocephaly..."	Macrocephaly
				Table 1: "...Gross motor delay..."	Mental deficiency
	B		Case: "...11 Maries et al. [1995] patient IV..."	Table 1: "...Brachydactyly..."	Brachydactyly
				Table 1: "...CAVC..."	Congenital heart defect
				Table 1: "...Ocular hypertelorism..."	Hypertelorism
				Table 1: "...Large anterior fontanelle..."	Large fontanelle
				Table 1: "...Short neck..."	Short neck
				Table 1: "...Syndactyly..."	Syndactyly
Craniocectodermal dysplasia	A	Amar MJ, Sutphen R, Kousseff BG. Expanded phenotype of craniocectodermal dysplasia (Sensenbrenner syndrome). Am J Med Genet. 1997 Jun 27;70(4):349–52.	Case: "...5: 7..."	Table 1: "...Brachydactyly..."	Brachydactyly
				Table 1: "...Sagittal suture synostosis..."	Craniosynostosis
				Table 1: "...Dental abnormalities..."	Dental anomaly
				Table 1: "...Dolichocephaly..."	Dolichocephaly
				Table 1: "...Epicanthal folds..."	Epicanthus
				Table 1: "...Frontal bossing..."	Frontal bossing
				Table 1: "...Hypotelorism..."	Hypotelorism
				Table 1: "...Rhizomelia..."	Rhizomelic limb
				Table 1: "...Short/narrow thorax..."	Small thorax
				Table 1: "...Syndactyly 2nd-3rd toes..."	Syndactyly
	B		Case: "...The reported patient..."	"...symmetric short fingers and toes were noted..."	Brachydactyly
				"...4th and 5th finger clinodactyly was present...."	Clinodactyly
				"...bone age was delayed, and no ossification centers were visible in hands, feet, hips and pelvis..."	Delayed osseous maturation
				"...two partially erupted, cone-shaped, lateral incisors..."	Dental anomaly

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
				"...mild dolichocephaly..."	Dolichocephaly
				Table 1: "...Hypertelorism..."	Hypertelorism
				"...head circumference (OFC) was 30.5 cm (3 SD below the mean)..."	Microcephaly
				"...At age 14 months the length, weight, and OFC were still 6 SD below the mean..."	Short stature
				"...simian creases..."	Simian crease
				"...mild cutaneous syndactyly of 2nd and 3rd toes..."	Syndactyly
Distal arthrogryposis	A	Gripp KW, Scott CI, Brockett BC, Nicholson L, Mackenzie WG. Extending the spectrum of distal arthrogryposis. American Journal of Medical Genetics. Wiley Subscription Services, Inc., A Wiley Company; 1996 Nov 11;65(4):286–90.	Case: "...Patient 1..."	...arachnodactyly...	Arachnodactyly
				"...camptodactyly of all four fingers bilaterlly and ..."	Camptodactyly
				"...Mild residual clubfeet with varus deformities of the hindfeet were noted."	Club foot
				...Flexion contractures of hips nd knees,...	Joint contracture
				...Ears were apparently low-set...	Low-set ear
				...Severe kyphoscoliosis developed during adolescence...	Scoliosis
				...Blepharophimosis was obvious...	Short palpebral fissure
				...height was 145.9cm (<3rdcentile; 50th centile for 11 1/2 years...	Short stature
				Fig. 2: "...microstomia,..."	Small mouth
				...ulnar deviation of the wrists were described at 3month...	Ulnar deviation of the hand
	B		Case: "...Patient 2..."	...camptodactyly,...	Camptodactyly
				...cleft in soft and hard palate...	Cleft palate
				...severe clubfoot deformities...	Club foot
				...Delayed ossification of sternum, pelvis, femur, tibia, and calcaneus were noted on skeletal radiographs;...	Delayed osseous maturation
				...Hirsutism involved arms and shins...	Hirsutism
				...Ulnar deviation and flexion contractures of wrists and single palmar flexion creases were seen bilaterally...	Joint contracture
				...ears were low-set...	Low-set ear
				Table 1: "...blepharophimosis,..."	Short palpebral fissure
				...mouth was small with limited opening...	Small mouth
				...Ulnar deviation and flexion contractures of wrists and single palmar flexion creases were seen bilaterally...	Ulnar deviation of the hand
EEC	A	Maas SM, de Jong TPVM, Buss P, Hennekam RCM. EEC syndrome and genitourinary anomalies: An update. American Journal of Medical Genetics. Wiley Subscription Services, Inc., A Wiley Company; 1996 Jun 14;63(3):472–8.	Case: "...Patient IV-2..."	Table 1: "...Dental anomalies..."	Dental anomaly
				Table 1: "...Ectrodactyly hand..."	Ectrodactyly
				Table 1: "...Onychodysplasia..."	Nail dysplasia
				Table 1: "...Syndactyly foot..."	Syndactyly
	B		Case: "...Patient V-3..."	Table 1: "...Ulnar deviation of third finger..."	Clinodactyly
				Table 1: "...Dental anomalies..."	Dental anomaly
				Table 1: "...Hypohidrosis..."	Hypohidrosis
				Table 1: "...Onychodysplasia..."	Nail dysplasia
				Table 1: "...Syndactyly foot..."	Syndactyly
Fibrodysplasia ossificans congenita	A	Liu H, Sawyer SL, Gos M, Grynspan D, Issa K, Ramphal R, et al. Atypical fibrodysplasia ossificans progressiva diagnosed by whole-exome sequencing. Am J Med Genet. 2015 Apr 21;167(6):1337–41.	Case: "...The infant..."	...uterine didelphys....	Bicornuate uterus
				...bilateral fifth finger clinodactyly...	Clinodactyly
				...limited range of motion in the left knee....	Knee limitation
				...polyhydramnios...	Polyhydramnios
				...intrauterine growth retardation...	Prenatal onset growth deficiency
				...proximally placed thumbs...	Proximal thumb
				...scoliosis of 19 degrees...	Scoliosis
				...left orbital mass, causing severe restrictive strabismus....	Strabismus
				...agenesis of the left thyroid lobe...	Thyroid anomaly
	B	Kaplan FS, Kobori JA, Orellana C, Calvo I, Rosello M, Martinez F, et al. Multi-system involvement in a severe variant of fibrodysplasia ossificans progressiva (ACVR1c.772G>A; R258G): A report of two patients. Am J Med Genet. 2015 Jun 11;167(10):2265–71.	Case: "...Patient 1..."	"...widespread areas of heterotopic ossification throughout the neck, back, and shoulders..."	Accessory ossification center
				"...no nails..."	Congenital missing nail
				"...sensorineural hearing loss..."	Deafness
				"...small malformed teeth..."	Dental anomaly
				"...low-set dysmorphic ears..."	Ear anomaly
				"...shunted hydroceph- alus,..."	Hydrocephalus
				"...macrocephaly..."	Macrocephaly
				"...microretrognathia..."	Micrognathia
				"...decreased range of motion of the shoulders, elbows and hips..."	Shoulder limitation
				"...dysconjugate gaze..."	Strabismus
Floating-Harbor	A	Hersh JH, Groom KR, Yen FF, Verdi GD. Changing phenotype in Floating-Harbor syndrome. Am J Med Genet. 1998 Feb 26;76(1):58–61.	Case: "...Present case..."	Table 1: "...e. Bulbous nose..."	Bulbous nose
				Table 1: "...c. 5th finger clinodactyly..."	Clinodactyly
				Table 1: "...d. Delayed bone age..."	Delayed osseous maturation
				Table 1: "...d. Posteriorly angulated ears..."	Ear anomaly
				Table 1: "...f. Speech delay..."	Mental deficiency
				Table 1: "...b. Short neck..."	Short neck
				Table 1: "...g. Short philtrum..."	Short philtrum
				Table 1: "...c. Short stature..."	Short stature
				Table 1: "...g. Strabismus..."	Strabismus
				Table 1: "...a. Triangular face..."	Triangular facies
	B	Lazebnik N, McPherson E, Rittmeyer LJ, Mulvihill JJ. The floating harbor syndrome with cardiac septal defect. Am J Med Genet. Wiley Subscription Services, Inc., A Wiley Company; 1996 Dec 18;66(3):300–2.	Case: "... Chudley et al. 1991..."	Table 1: "...Clinodactyly..."	Clinodactyly
				Table 1: "...Deep set eyes..."	Deep-set eye
				Table 1: "...Posteriorly rotated ears..."	Ear anomaly
				Table 1: "...Delayed motor skills..."	Mental deficiency
				Table 1: "...Short stature < 2 SD..."	Short stature

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
				Table 1: "...Thin lips..."	Thin lip
				Table 1: "...Triangular face..."	Triangular facies
Gapo	A	Goloni-Bertollo EM, Ruiz MT, Goloni CBV, Muniz MP, Valério NI, Pavarino-Bertelli ÉC. GAPO syndrome: Three new Brazilian cases, additional osseous manifestations, and review of the literature. Am J Med Genet. Wiley Subscription Services, Inc., A Wiley Company; 2008;146A(12):1523–9.	Case: "...Wajntal et al. 1990: 2, M..."	Table 1: "...Alopecia..."	Alopecia
				Table 1: "...Delayed bone maturation..."	Delayed osseous maturation
				Table 1: "...Protruding auricles..."	Ear cartilage hypertrophy
				Table 1: "...Bossed forehead..."	Frontal bossing
				Table 1: "...Hepatomegaly..."	Hepatomegaly
				Table 1: "...Bilateral keratocomus..."	Keratoconus
				Table 1: "...Depressed nasal bridge..."	Low nasal bridge
				Table 1: "...Micrognathia..."	Micrognathia
				Table 1: "...Alteration of the nails..."	Nail dysplasia
				Table 1: "...Growth retardation..."	Prenatal onset growth deficiency
	B		Case: "...Sayli and Gül 1993: 1, M..."	Table 1: "...Alopecia..."	Alopecia
				Table 1: "...Bossed forehead..."	Frontal bossing
				Table 1: "...Hyperextensible joints..."	Joint hypermobility
				Table 1: "...Micrognathia..."	Micrognathia
				Table 1: "...Alteration of the nails..."	Nail dysplasia
				Table 1: "...Growth retardation..."	Prenatal onset growth deficiency
				Table 1: "...Protruding lips..."	Prominent lip
				Table 1: "...Skin redundancy..."	Redundand skin
Gillespie	A	Nelson J, Flaherty M, Grattan Smith P. Gillespie syndrome: A report of two further cases. American Journal of Medical Genetics. Wiley Subscription Services, Inc., A Wiley Company; 1997 Aug 8;71(2): 134–8.	Case: "...Patient 1..."	"...partial aniridia and pupillary membrane remnants were noted...."	Aniridia
				"...was ataxic..."	Ataxia
				"...hypotonic but strength was normal...."	Hypotony
				"...Wisps of pupillary membrane were also evident and the remaining iris was hypoplastic..."	Iris hypoplasia
				"...motor development was delayed..."	Mental deficiency
				"...On examination at 4 7/12 years her height was on the 75th centile, weight was on the 50th centile, and head circumference (OFC) was below the 98th centile...."	Microcephaly
				"...An intermittent horizontal and at times vertical nystagmus was present with an exotropia...."	Nystagmus
				"...Spinal X-ray films showed partial fusion involving cervical vertebrae 2 and 3 with mild hypoplasia of the odontoid (Fig. 1D)...."	Vertebral anomaly
	B		Case: "...Case 2..."	"...partial aniridia..."	Aniridia
				"...very ataxic..."	Ataxia
				"...A right inguinal hernia repair and right orchidopexy were performed at 6 weeks...."	Cryptorchidism
				"...high frequency hearing loss..."	Deafness
				"...markedly hypotonic..."	Hypotony
				"...A right inguinal hernia repair and right orchidopexy were performed at 6 weeks...."	Inguinal hernia
				"...lamp examination showed bilateral symmetrical iris hypoplasia...."	Iris hypoplasia
				"...have bilateral ptosis,..."	Ptosis of eyelid
				"...failure to thrive due t..."	Short stature
				"...upslanting palpebral fissures..."	Upward slanted palpebral fissure
Holt-Oram	A	Sletten LJ, Pierpont ME. Variation in severity of cardiac disease in Holt-Oram syndrome. Am J Med Genet. Wiley Subscription Services, Inc., A Wiley Company; 1996 Oct 16;65(2):128–32.	Case: "...Patient 2 (III-7)..."	"...large secundum ASD..."	Atrial septum defect
				"...Evaluation here at 22 years showed the presence of triphalangeal thumbs with de creased thenar musculature (Fig...."	Triphalangeal thumb
				"...small VSD..."	Ventricular septum defect
	B	Morine M, Kohmoto T, Masuda K, Inagaki H, Watanabe M, Naruto T, et al. A unique TBX5microdeletion with microinsertion detected in patient with Holt-Oram syndrome. Am J Med Genet. 2015 Sep 28;167(12):3192–6.	Case: "...CLINICAL REPORT..."	...multiple muscular and perimembranous ASD and VSD...	Atrial septum defect
				...the right upper limb had a radially bowed forearm...	Bowed bone
				...right folded ear...	Ear anomaly
				...with flexion deformity at the elbow and wrist joint...	Elbow dysplasia
				...persistent left superior vena cava...	Left superior vena cava
				...hypoplasia of the left humerus...	Short humerus
				...multiple muscular and perimembranous ASD and VSD...	Ventricular septum defect
Johnson-mcmillin	A	Schweitzer DN, Yano S, Earl DL, Graham JM. Johnson-McMillin syndrome, a neuroectodermal syndrome with conductive hearing loss and microtia: Report of a new case. Am J Med Genet. Wiley Subscription Services, Inc., A Wiley Company; 2003 Jun 26;120A(3):400–5.	Case: "...Our case: P..."	Table 1: "...Alopecia..."	Alopecia
				Table 1: "...Cleft palate..."	Cleft palate
				Table 1: "...Congenital heart defectd..."	Congenital heart defect
				Table 1: "...Conductive hearing loss..."	Deafness
				Table 1: "...Ear abnormality..."	Ear anomaly
				Table 1: "...Facial asymmetry..."	Facial asymmetry
				Table 1: "...Mental retardation..."	Mental deficiency
				Table 1: "...microcephaly..."	Microcephaly
				Table 1: "...Growth deficiency..."	Prenatal onset growth deficiency
	B		Case: "...Johnston et al. [1987]..."	Table 1: "...Alopecia..."	Alopecia
				Table 1: "...Cafe-au-lait spots..."	Cafe-au-lait spot
				Table 1: "...Ear abnormality..."	Ear anomaly
				Table 1: "...Mental retardation..."	Mental deficiency
				Table 1: "...microcephaly..."	Microcephaly
Kabuki make-up	A	Tsukahara M, Kuroki Y, Imaizumi K, Miyazawa Y, Matsuo K. Dominant inheritance of Kabuki make-up syndrome. Am J Med Genet. 1997 Nov 28;73(1):19–23.	Case: "...Family 1: Mother 27..."	Table 1: "...Cleft palate..."	Cleft palate
				Table 1: "...Lower palpebral eversion..."	Ectropion
				Table 1: "...Epicanthus..."	Epicanthus
				Table 1: "...Arched eyebrows, sparse in lateral half..."	High arched eyebrow

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
				Table 1: "...IQ or mental retardation..."	Mental deficiency
				Table 1: "...Prominent ears..."	Prominent ear
				Table 1: "...Short stature..."	Short stature
				Table 1: "...Strabismus..."	Strabismus
	B	Karagianni P, Lambropoulos V, Stergidou D, Fryssira H, Chatziioannidis I, Spyridakis I. Recurrent giant cell fibroblastoma: Malignancy predisposition in Kabuki syndrome revisited. Am J Med Genet. 2016 Feb 22;170(5):1333–8.	Case: "...Our female patient..."	...sinus venosus atrial septal defect...	Atrial septum defect
				...uvula bifida...	Bifid uvula
				...mild dilation of lateral ventricles...	Congenital heart defect
				...ectopic pelvic kidneys...	Ectopic kidney
				...epicanthus...	Epicanthus
				...multiple middle ear and upper respira- tory tract infections...	Immunity deficiency
				...neurodevelopmental delay...	Mental deficiency
				...micrognathia...	Micrognathia
				...transverse position of the right kidney which was located near the middle line...	Renal anomaly
				...Precocious puberty...	Sexual precocity
Karsch-Neugebauer	A	Wong SC, Cobben JM, Hiemstra S, Robinson PH, Heeg M. Karsch-Neugebauer syndrome in two sibs with unaffected parents. Am J Med Genet. 1998 Jan 13;75(2):207–10.	Case: "...Karsch [1936] (1)..."	Table 1: "...Cataract/ retinal abnormalities..."	Cataract
				Table 1: "...Split hand..."	Ectrodactyly
				Table 1: "...Nystagmus..."	Nystagmus
	B		Case: "...Pilarski et al. [1985] (1)..."	Table 1: "...Split hand..."	Ectrodactyly
				Table 1: "...Nystagmus..."	Nystagmus
Kaufman oculo-cerebro-facial	A	Pedurupillay CRJ, Barøy T, Holmgren A, Blomhoff A, Vigeland MD, Sheng Y, et al. Kaufman oculocerebrofacial syndrome in sisters with novel compound heterozygous mutation in UBE3B. Am J Med Genet. 2015 Feb 18;167(3):657–63.	Case: "...Patient 1..."	...increased posterior angulation of the ears...	Ear anomaly
				...epicanthus...	Epicanthus
				...thick and highly arched eyebrows with cleft...	High arched eyebrow
				...widely spaced eyes...	Hypertelorism
				...globally delayed and slightly growth retarded...	Mental deficiency
				...OFC 29.5 cm (1 cm <3rd centile)...	Microcephaly
				Table 1: "...Nail dysplasia..."	Nail dysplasia
				Table 1: "...Ptosis..."	Ptosis of eyelid
				...height 93 cm (3 cm <3rd centile)...	Short stature
				...upslanting palpebral fissures...	Upward slanted palpebral fissure
	B		Case: "...Patient 2..."	...Genitalia were ambiguous...	Ambiguous genitalia
				...cleft uvula...	Bifid uvula
				...a submucous cleft palate...	Cleft palate
				...increased posterior angulation of the ears...	Ear anomaly
				...epicanthus...	Epicanthus
				...thick and highly arched eyebrows with cleft...	High arched eyebrow
				...life), widely spaced eyes...	Hypertelorism
				...severe intellectual disability...	Mental deficiency
				...OFC 31 cm (1 cm <3rd centile...	Microcephaly
				...Birth weight was 2,130 g (300 g <3rd centile)...	Prenatal onset growth deficiency
Kenny-Caffey	A	Tahseen K, Khan S, Uma R, Usha R, Ghanem Al MM, Awadi Al SA, et al. Kenny-Caffey syndrome in six Bedouin sibships: autosomal recessive inheritance is confirmed. Am J Med Genet. 1997 Mar 17;69(2):126–32.	Case: "...B: II-10..."	Table 1: "...Cortical thickening and medullary stenosis of long bones..."	Bone cortical thickening
				Table 1: "...Hypocalcemia..."	Hypocalcemia
				Table 1: "...Psychomotor retardation..."	Mental deficiency
				Table 1: "...Birth weight < 2.5 kg..."	Prenatal onset growth deficiency
				Table 1: "...Dwarfism..."	Short stature
				Table 1: "...Absent diploic space in calvaria..."	Thin calvarium
	B		Case: "...B: II-7..."	Table 1: "...Cortical thickening and medullary stenosis of long bones..."	Bone cortical thickening
				Table 1: "...Dental caries..."	Caries
				Table 1: "...Delayed bone age..."	Delayed osseous maturation
				Table 1: "...Hypocalcemia..."	Hypocalcemia
				Table 1: "...Psychomotor retardation..."	Mental deficiency
				Table 1: "...Birth weight < 2.5 kg..."	Prenatal onset growth deficiency
				Table 1: "...Dwarfism..."	Short stature
				Table 1: "...Absent diploic space in calvaria..."	Thin calvarium
Keutel	A	Teebi AS, Lambert DM, Kaye GM, Al-Fifi S, Tewfik TL, Azouz EM. Keutel syndrome: further characterization and review. Am J Med Genet. 1998 Jun 30;78(2):182–7.	Case: "...Zieriesen et al., 1993..."	Table 1: "...Brachytelephalangism..."	Brachydactyly
				Table 1: "...Peripheral pulmonary stenosis/heart defects..."	Congenital heart defect
				Table 1: "...Borderline/Mild MR..."	Mental deficiency
				Table 1: "...Peripheral pulmonary stenosis/heart defects..."	Peripheral pulmonary arterial stenosis
				Table 1: "...Abnormal cartilage calcification..."	Synostosis
	B	Weaver KN, Hallek El M, Hopkin RJ, Sund KL, Henrickson M, del Gaudio D, et al. Keutel syndrome: Report of two novel MGPmutations and discussion of clinical overlap with arylsulfatase E deficiency and relapsing polychondritis. Am J Med Genet. 2014 Jan 23;164(4):1062–8.	Case: "...Patient 3..."	"...Regional alopecia was observed..."	Alopecia
				"...moderate sensorineural hearing loss (43 dB) in the right ear and severe mixed hearing loss (67 dB) in the left ear...."	Deafness
				"...auricular cartilages were stiff and slightly posteriorly angulated..."	Ear anomaly
				"...calcification of auricular, nasal septum, laryn- geal, and tracheal cartilages...."	Ectopic calcification
				"...Her fingers had a drumstick appearance..."	Finger sausage-like
				"...Dental malocclusion..."	Irregular placement of teeth
				"...depressed nasal bridge..."	Low nasal bridge
				"...wide mouth..."	Macrostomia
				"...short distal phalanges..."	Phalangeal distal hypoplasia
				"...short philtrum..."	Short philtrum

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
Knobloch	A	Wilson C, Aftimos S, Pereira A, McKay R. Report of two sibs with Knobloch syndrome (encephalocoele and viteoretinal degeneration) and other anomalies. American Journal of Medical Genetics. John Wiley & Sons, Inc; 1998 Jul 7;78(3):286–90.	Case: "...Case 1..."	"...dolichocephalic skull..."	Dolichocephaly
				"...mild hepatomegaly..."	Hepatomegaly
				"...mild micrognathia..."	Micrognathia
				"...Initial ophthalmological opinion showed he had an alternating convergent squint and was slightly myopic..."	Myopia
				"...and a small patent ductus arteriosus (PDA)..."	Patent ductus arteriosus
				"...An abdominal ultrasound study on day 30 showed pyloric stenosis..."	Pyloric stenosis
				"...single umbilical artery..."	Single umbilical artery
				"...strabismus..."	Strabismus
				"...At 4 months he remained tachypneic..."	Tachypnoe
				"...small midmuscular ventricular septal defect (VSD..."	Ventricular septum defect
	B		Case: "...Case 2..."	"...dolichocephalic skul..."	Dolichocephaly
				"...lop ears similar to those of his brother..."	Ear anomaly
				"...These findings were consistent with an encephalocele..."	Encephalocele
				Table 1: "...Myopia..."	Myopia
				"...coarse nystagmus...."	Nystagmus
				"...showed the right eye to have some posterior subcapsular lens opacity and peripheral vitreoretinopathy with an abnormal disc and dragging of the retina inferiorly. There was also some lens opacity in the left eye, and advanced vitreoretinopathy had led to total retinal detachment with subretinal hemorrhage (Fig. 3)...."	Retinal detachment
				"...Clinical review at 3 months showed him to be still tachypneic but otherwise clinically well..."	Tachypnoe
Malpuech	A	Crisponi G, Marras AR, Corrias A. Two sibs with Malpuech syndrome. American Journal of Medical Genetics. Wiley Subscription Services, Inc., A Wiley Company; 1999 Sep 17;86(3): 294–9.	Case: "...Our patients: 2..."	Table 1: "...Wide forehead..."	Broad forehead
				Table 1: "...Cleft lip/palate..."	Cleft lip
				Table 1: "...Cleft lip/palate..."	Cleft palate
				Table 1: "...Hearing loss..."	Deafness
				Table 1: "...Hypertelorism..."	Hypertelorism
				Table 1: "...Inguinal hernia..."	Inguinal hernia
				Table 1: "...Malar hypoplasia..."	Micrognathia
				Table 1: "...Growth retardation..."	Prenatal onset growth deficiency
				Table 1: "...Ptosis..."	Ptosis of eyelid
				Table 1: "...Omphalocele/wide umbilical hernia..."	Umbilical hernia
	B		Case: "...Our patients: 1..."	Table 1: "...Wide forehead..."	Broad forehead
				Table 1: "...Cleft lip/palate..."	Cleft lip
				Table 1: "...Cleft lip/palate..."	Cleft palate
				Table 1: "...Hearing loss..."	Deafness
				Table 1: "...Arched eyebrows..."	High arched eyebrow
				Table 1: "...Hypertelorism..."	Hypertelorism
				Table 1: "...Malar hypoplasia..."	Micrognathia
				Table 1: "...Growth retardation..."	Prenatal onset growth deficiency
				Table 1: "...Ptosis..."	Ptosis of eyelid
Menkes	A	Smpokou P, Samanta M, Berry GT, Hecht L, Engle EC, Lichter-Konecki U. Menkes disease in affected females: the clinical disease spectrum. Am J Med Genet. 2015 Feb;167A(2):417–20.	Case: "...Patient 3..."	"...truncal ataxia..."	Ataxia
				"...Cerebellar atrophy..."	Cerebral atrophy
				"...mitral valve prolapse with mild mitral regurgitation and severe aortic root dilation..."	Congenital heart defect
				"...hypotonia..."	Hypotony
				"...skin and joint laxity..."	Joint hypermobility
				"...poor growth and developmental delay..."	Mental deficiency
				"...epilepsy..."	Seizure
				"...wormian" skull bones..."	Wormian bone
	B	Burgemeister AL, Zirn B, Oeffner F, Kaler SG, Lemm G, Rossier E, et al. Menkes disease with discordant phenotype in female monozygotic twins. Am J Med Genet. 2015 Aug 4;167(11):2826–9.	Case: "...The MZT girls..."	...ataxia...	Ataxia
				...bladder diverticulae....	Bladder diverticulum
				...progressive brain atrophy...	Cerebral atrophy
				...severe hypotonia...	Hypotony
				...recurrent infections,...	Immunity deficiency
				...inguinal hernia was diagnosed...	Inguinal hernia
				...hypopigmented kinky hair...	Kinky hair
				...almost no motor development...	Mental deficiency
				...nystagmus with headnodding...	Nystagmus
Multiple lentigines	A	Digilio MC, Sarkozy A, de Zorzi A, Pacileo G, Limongelli G, Mingarelli R, et al. LEOPARD syndrome: Clinical diagnosis in the first year of life. Am J Med Genet. 2006;140A(7):740–6.	Case: "...Patient 5, M..."	Table 1: "...Cafe-au-lait spots..."	Cafe-au-lait spot
				Table 1: "...HCM..."	Cardiomyopathy
				Table 1: "...CHD..."	Congenital heart defect
				Table 1: "... Downs slanting palpebral fissures..."	Downward slanted palpebral fissure
				Table 1: "...Dysmorphic ears..."	Ear anomaly
				Table 1: "...Hypertelorism..."	Hypertelorism
				Table 1: "...Delayed milestones..."	Mental deficiency
				Table 1: "...Weight <3rd centile..."	Prenatal onset growth deficiency
				Table 1: "...Ptosis..."	Ptosis of eyelid
				Table 1: "...Length <3rd centile..."	Short stature
	B		Case: "...Patient 3, F..."	Table 1: "...ASD..."	Atrial septum defect
				Table 1: "...Cafe-au-lait spots..."	Cafe-au-lait spot
				Table 1: "...HCM..."	Cardiomyopathy
				Table 1: "...CHD..."	Congenital heart defect
				Table 1: "...Dysmorphic ears..."	Ear anomaly
				Table 1: "...Hypertelorism..."	Hypertelorism

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
				Table 1: "...Low-set ears with overfolded helix..."	Low-set ear
				Table 1: "...Short neck..."	Short neck
Mutchinick	A	Doerfler W, Wieczorek D, Gillessen-Kaesbach G, Albrecht B, Passarge E. Three brothers with mental and physical retardation, hydrocephalus, microcephaly, internal malformations, speech disorder, and facial anomalies: Mutchinick syndrome. Am J Med Genet. 1997 Dec 12;73(2):210–6.	Case: "...This report: M.K. VI-4..."	Table 1: "...Light blue iris..."	Blue iris
				Table 1: "...Frequent caries..."	Caries
				Table 1: "...Antimongoloid palpebral fissures..."	Downward slanted palpebral fissure
				Table 1: "...Large, protruding low-set, dysplastic ears..."	Ear anomaly
				Table 1: "...Hydrocephalus..."	Hydrocephalus
				Table 1: "...Large mouth..."	Macrostomia
				Table 1: "...microcephaly..."	Microcephaly
				Table 1: "...Prognathism..."	Prognathism
				Table 1: "...Prominent nose and nasal bridge..."	Prominent nose
				Table 1: "...Ptosis..."	Ptosis of eyelid
	B		Case: "...This report: Mat. K. VI-2..."	Table 1: "...Light blue iris..."	Blue iris
				Table 1: "...High forehead..."	Broad forehead
				Table 1: "...Irregular ridges on incisors..."	Dental anomaly
				Table 1: "...Antimongoloid palpebral fissures..."	Downward slanted palpebral fissure
				Table 1: "...Large, protruding low-set, dysplastic ears..."	Ear anomaly
				Table 1: "...Large mouth..."	Macrostomia
				Table 1: "...microcephaly..."	Microcephaly
				Table 1: "...Prognathism..."	Prognathism
				Table 1: "...Prominent nose and nasal bridge..."	Prominent nose
				Table 1: "...Ptosis..."	Ptosis of eyelid
Myhre	A	Starr LJ, Grange DK, Delaney JW, Yetman AT, Hammel JM, Sanmann JN, et al. Myhre syndrome: Clinical features and restrictive cardiopulmonary complications. Am J Med Genet. 2015 Sep 30;167(12):2893–901.	Case: "...Patient 2..."	"...mild coarctation of the aorta..."	Coarctation of aorta
				"...superimposed bilateral sensorineural hearing loss resulting in severe speech delay..."	Deafness
				"...prenatally diagnosed duodenal atresia..."	Duodenal atresia
				...broad ears with a squared shape...	Ear anomaly
				...midfacial retrusion...	Midfacial hypoplasia
				"...polyhydramnios..."	Polyhydramnios
				...prognathism...	Prognathism
				"...11 rib pairs..."	Rib anomaly
				"...persistent short stature..."	Short neck
				...microstomia...	Small mouth
	B		Case: "...Patient 4..."	...Mild brachydactyly...	Brachydactyly
				"...two years of age bilateral hearing loss..."	Deafness
				"...hypotonia..."	Hypotony
				"...restricted finger joint movement..."	Joint limitation
				"...motor development was mildly delayed..."	Mental deficiency
				...mild midfacial retrusion...	Midfacial hypoplasia
				...relative prognathism...	Prognathism
				"...short stature..."	Short stature
Neu-laxova	A	Mattos EP, Silva AAD, Magalhães JAA, Leite JCL, Leistner-Segal S, Gus-Kessler R, et al. Identification of a premature stop codon mutation in the PHGDHgene in severe Neu-Laxova syndrome-evidence for phenotypic variability. Am J Med Genet. 2015 Apr 25;167(6):1323–9.	Case: "Result"	...club feet...	Club foot
				...mild ichthyosis...	Ichthyosis
				...with contractures of upper and lower limbs...	Joint contracture
				...absence of cerebral gyri and sulci reinforced the prenatal finding of lissencephaly....	Lissencephaly
				...microcephaly...	Microcephaly
				...micrognathia...	Micrognathia
				...symmetric growth restriction,...	Prenatal onset growth deficiency
				...shortened limbs...	Short limb
				...short neck...	Short neck
				...spina bifida...	Spina bifida
	B	Hickey P, Piantanida E, Lentz-Kapua S, Kenner J. Neu-Laxova syndrome: a case report. Pediatr Dermatol. 2003 Jan;20(1):25–7.	Case: "...A Caucasian infant..."	"...dilated ventricles,..."	Cardiomyopathy
				"...hydrocephalus ex vacuo...."	Hydrocephalus
				"...Despite excellent home nursing, the child's outpatient course was complicated by frequent candidiasis of the skin and eyes, pseudomonal skin and urinary tract infections, and corynebacteria bacteremia...."	Immunity deficiency
				"...limb contractures..."	Joint contracture
				"...A head computed tomography (CT) scan revealed partial lissencephaly (absence of gyri)..."	Lissencephaly
				"...Additional abnormal clinical findings included microcephaly..."	Microcephaly
				"...growth retardation..."	Prenatal onset growth deficiency
				"...short neck..."	Short neck
				"...sloping forehead..."	Sloping forehead
Noonan	A	Addissie YA, Kotecha U, Hart RA, Martinez AF, Kruszka P, Muenke M. Craniosynostosis and Noonan syndrome with KRASmutations: Expanding the phenotype with a case report and review of the literature. Am J Med Genet. 2015 Aug 6;167(11):2657–63.	Case: "...The proband..."	...hair was sparse around the fronto- temporal region...	Alopecia
				...tall forehead...	Broad forehead
				...have broad great toes suggestive of a cranio- synostosis syndrome...	Broad toe
				...sagittal suture synostosis...	Craniosynostosis
				...left down slanting palpebral fissure...	Downward slanted palpebral fissure
				...thick helices....	Ear cartilage hypertrophy
				...hypertelorism...	Hypertelorism
				...low-set ears...	Low-set ear
				...bilateral ptosis...	Ptosis of eyelid
				Table 1: "...Shortstature..."	Short stature

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
	B	Guerin A, So J, Mireskandari K, Jougeh-Doust S, Chisholm C, Klatt R, et al. Expanding the clinical spectrum of ocular anomalies in Noonan syndrome: Axenfeld-anomaly in a child with PTPN11mutation. Am J Med Genet. 2014 Nov 25;167(2):403–6.	Case: *...Our patient..."	*...with hypertrophic cardiomyopathy..."	Cardiomyopathy
				*...head and face were edematous..."	Cheek edema
				*...dental hypo- plasia,..."	Dental anomaly
				*...downslanted palpebral fissures..."	Downward slanted palpebral fissure
				*...Axenfeld anomaly with glaucoma in left eye..."	Glaucoma
				*...hypertelorism..."	Hypertelorism
				*...depressed nasal bridge..."	Low nasal bridge
				*...posteriorly rotated, low-set ears...."	Low-set ear
				*...head circum- ference of 43.3 cm (at the 2nd centile, which was thought to be secondary to perinatal complications)..."	Microcephaly
				*...revealed a patent foramen ovale and a patent ductus arteriosus...."	Patent ductus arteriosus
Opitz-kaveggia	A	Graham JM, Superneau D, Rogers RC, Corning K, Schwartz CE, Dykens EM. Clinical and behavioral characteristics in FG syndrome. Am J Med Genet. 1999 Aug 27;85(5):470–5.	Case: *...Graham et al. [1998]: 5..."	Table 1: *...Agenesis corpus callosum..."	Agenesis of corpus callosum
				Table 1: *...Tall, broad forehead..."	Broad forehead
				Table 1: *...Broad thumbs/halluces..."	Broad thumb
				Table 1: *...Broad thumbs/halluces..."	Broad toe
				Table 1: *...Abnormal auricles..."	Ear anomaly
				Table 1: *...Ocular hypertelorism..."	Hypertelorism
				Table 1: *...Congenital hypotonia..."	Hypotony
				Table 1: *...Mental retardation..."	Mental deficiency
	B		Case: *...Graham et al. [1998]: 1..."	Table 1: *...Agenesis corpus callosum..."	Agenesis of corpus callosum
				Table 1: *...Tall, broad forehead..."	Broad forehead
				Table 1: *...Cryptorchidism..."	Cryptorchidism
				Table 1: *...Abnormal auricles..."	Ear anomaly
				Table 1: *...Ocular hypertelorism..."	Hypertelorism
				Table 1: *...Congenital hypotonia..."	Hypotony
				Table 1: *...Mental retardation..."	Mental deficiency
				Table 1: *...Scoliosis..."	Scoliosis
				Table 1: *...Seizures/abnormal EEG..."	Seizure
				Table 1: *...Single palm crease..."	Simian crease
Osteoglophonic	A	Sklower Brooks S, Kassner G, Qazi Q, Keogh MJ, Gorlin RJ. Osteoglophonic dysplasia: review and further delineation of the syndrome. American Journal of Medical Genetics. Wiley Subscription Services, Inc., A Wiley Company; 1996 Dec 11;66(2): 154–62.	Case: *...Stoß et al., 1991..."	Table II: *...Cystic metaphyseal defects..."	Bone metaphyseal lesion
				Table II: *...Abnormal skull shape at birth..."	Cranial asymmetry
				Table II: *...Craniostenosis..."	Craniosynostosis
				Table II: *...Frontal bossing..."	Frontal bossing
				Table II: *...Hypertelorism..."	Hypertelorism
				Table II: *...Platyspondyly..."	Platyspondyly
				Table II: *...1,840 g..."	Prenatal onset growth deficiency
				Table II: *...Rhizomelic dwarfism..."	Rhizomelic limb
				Table II: *...Failure to thrive..."	Short stature
	B		Case: *...Santos et al., 1988..."	Table II: *...Anteverted nares..."	Anteverted nare
				Table II: *...Cystic metaphyseal defects..."	Bone metaphyseal lesion
				Table II: *...Abnormal skull shape at birth..."	Cranial asymmetry
				Table II: *...Craniostenosis..."	Craniosynostosis
				Table II: *...Frontal bossing..."	Frontal bossing
				Table II: *...Hypertelorism..."	Hypertelorism
				Table II: *...Unerupted teeth..."	Late eruption of teeth
				Table II: *...Platyspondyly..."	Platyspondyly
				Table II: *...Prognathism..."	Prognathism
				Table II: *...Rhizomelic dwarfism..."	Rhizomelic limb
Perlman	A	Henneveld HT, van Lingen RA, Hamel BC, Stolte-Dijkstra I, van Essen AJ. Perlman syndrome: four additional cases and review. Am J Med Genet. 1999 Oct 29;86(5):439–46.	Case: *...Liban et al., 1970; Perlman et al., 1973; Perlman et al., 1975: Patient 5..."	Table 1: *...Broad, depressed nasal bridge..."	Broad nasal bridge
				Table 1: *...Deep-set eyes..."	Deep-set eye
				Table 1: *...Hepatomegaly..."	Hepatomegaly
				Table 1: *...Hypotonia..."	Hypotony
				Table 1: *...Low-set ears..."	Low-set ear
				Table 1: *...Macrocephaly (>97th centile) (cm)..."	Macrocephaly
				Table 1: *...Nephroblastomatosis..."	Nephroblastoma
				Table 1: *...Polyhydramnios..."	Polyhydramnios
				Table 1: *...Nephromegaly..."	Renal anomaly
				Table 1: *...Wilms tumor..."	Wilms tumor
	B		Case: *...This report: Patient A..."	Table 1: *...Epicanthal folds..."	Epicanthus
				Table 1: *...Hepatomegaly..."	Hepatomegaly
				Table 1: *...Hypoglycaemia..."	Hypoglycemia
				Table 1: *...Low-set ears..."	Low-set ear
				Table 1: *...Macrocephaly (>97th centile) (cm)..."	Macrocephaly
				Table 1: *...Micro-/retrognathia..."	Micrognathia
				Table 1: *...Nephroblastomatosis..."	Nephroblastoma
				Table 1: *...Polyhydramnios..."	Polyhydramnios
				Table 1: *...Nephromegaly..."	Renal anomaly
				Table 1: *...Wilms tumor..."	Wilms tumor
Pitt-Rogers-Danks	A	Clemens M, Martsolf JT, Rogers JG, Mowery Rushton P, Surti U, McPherson E. Pitt-Rogers-Dank syndrome: The result of a 4p microdeletion. American Journal of Medical Genetics. Wiley Subscription Services, Inc., A Wiley Company; 1996 Dec 2;66(1): 95–100.	Case: *...Pitt et al: D..."	Table 1: *...High forehead..."	Broad forehead
				Table 1: *...Ear anomalies..."	Ear anomaly
				Table 1: *...Hyperactivity..."	Hyperactivity
				Table 1: *...Hypertel/teleca..."	Hypertelorism

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
				Table 1: *...Wide mouth..."	Macrostomia
				Table 1: *...microcephaly..."	Microcephaly
				Table 1: *...Max hypoplasia..."	Micrognathia
				Table 1: *...IUGR..."	Prenatal onset growth deficiency
				Table 1: *...Epilepsy..."	Seizure
				Table 1: *...Short stature..."	Short stature
	B		Case: *...Lizcano 1995..."	Table 1: *...Beaked nose..."	Beaked nose
				Table 1: *...Ear anomalies..."	Ear anomaly
				Table 1: *...Hypertel/teleca..."	Hypertelorism
				Table 1: *...Wide mouth..."	Macrostomia
				Table 1: *...Dev delay..."	Mental deficiency
				Table 1: *...microcephaly..."	Microcephaly
				Table 1: *...Max hypoplasia..."	Micrognathia
				Table 1: *...IUGR..."	Prenatal onset growth deficiency
				Table 1: *...Short philtrum..."	Short philtrum
				Table 1: *...Short stature..."	Short stature
Prader-Willi	A	Morandi A, Bonnefond A, Lobbens S, Carotenuto M, del Giudice EM, Froguel P, et al. A girl with incomplete Prader-Willi syndrome and negative MS-PCR, found to have mosaic maternal UPD-15 at SNP array. Am J Med Genet. 2015 Jun 24;167(11):2720–6.	Case: *...CLINICAL REPORT..."	...except for bilaterally diadokokinesis and heel-knee test....	Ataxia
				...a global developmental delay...	Mental deficiency
				...early obesity...	Obesity
				...short stature (height 1/4 147 cm, -2.3 SD and 16 cm below the genetic target of 163 cm)...	Short stature
	B	Butler MG, Christian SL, Kubota T, Ledbetter DH. A 5-year-old white girl with Prader-Willi syndrome and a submicroscopic deletion of chromosome 15q11q13. Am J Med Genet. 1996 Oct 16;65(2):137–41.	Case: *...EE..."	*...downturned corners of the mouth..."	Downturning corners of mouth
				*...outer canthal distance 6.5 cm (<3rd centile)..."	Hypotelorism
				*...se vere hypotonia..."	Hypotony
				*...micrognathia..."	Micrognathia
				*...have failure to thrive..."	Short stature
				*...short upturned nose..."	Small nose
Robinow	A	Bunn KJ, Lai A, Al-Ani A, Farella M, Craw S, Robertson SP. An osteosclerotic form of Robinow syndrome. Am J Med Genet. 2014 Jul 14;164(10):2638–42.	Case: *...Patient 2..."	*...Brachydactyly..."	Brachydactyly
				*...Left cleft lip and palate..."	Cleft lip
				*...Left cleft lip and palate..."	Cleft palate
				*...Bilateral intra-abdominal testes were observed and surgically treated by left orchidectomy and right orchidopexy..."	Cryptorchidism
				*...none of his primary teeth exfoliated prompting their removal from the lower jaw at 11 years of age..."	Dental anomaly
				*...apparent hypertelorism..."	Hypertelorism
				*...dental crowding..."	Irregular placement of teeth
				*...When evaluated at 9 years of age the patient weighed 35.4 kg (90th–97th centile), had a height of 135 cm (75th–90th centile), and a head circumference of 62.7 cm (>97th centile..."	Macrocephaly
				*...impression of short limbs..."	Short limb
				*...umbilical hernia were noted at birth;..."	Umbilical hernia
	B		Case: *...Patient 1..."	*...generalized osteosclerosis affecting the skull, axial and appendicular skeleton..."	Bone sclerosis
				*...brachydactyly were also observed..."	Brachydactyly
				*...camptodactyly..."	Camptodactyly
				*...clinodactyly..."	Clinodactyly
				*...Bilateral mixed conductive and sensorineural hearing loss was diagnosed..."	Deafness
				*...agenesis of 12 secondary teeth...."	Dental anomaly
				*...down-slanting palpebral fissures..."	Downward slanted palpebral fissure
				*...hypertelorism (IC distance 4cm, >97th centile)..."	Hypertelorism
				*...Oral examination revealed moderate tooth crowding, severe mesial rotation of the upper left incisor..."	Irregular placement of teeth
				*...at age 13 years the patient weighed 40 kg (10th centile), had a height of 147.8cm (<10th centile), and a head circumference of 60 cm (>97th centile..."	Macrocephaly
Rubinstein-Taybi	A	Bedeschi MF, Crippa BL, Colombo L, Guez S, Cerruti M, Fogliani R, et al. Unusual prenatal presentation of Rubinstein-Taybi syndrome: A case report. Am J Med Genet. 2014 Jul 29;164(10): 2663–6.	Case: *...The propositus..."	*...agenesis of the posterior third of the corpus callosum..."	Agenesis of corpus callosum
				*...cataracts..."	Cataract
				Table 1: *...Dandy–Walker malformation..."	Dandy-walker malformation
				*...glaucoma..."	Glaucoma
				*...hypertelorism..."	Hypertelorism
				*...low-set ears..."	Low-set ear
				*...bilat- eral megalocornea..."	Megalocornea
				*...cranial circumference less than 3rd centile, persistence of the globally thinned corpus callosum with a resulting deformity of the lateral ventricles and bilateral malrotation of hippocampus...."	Microcephaly
				*...pre and post-axial bilateral polydactyly of feet..."	Polydactyly
				*...Polyhydramnios was detected at the 35th week...."	Polyhydramnios
	B	Solomon BD, Bodian DL, Khromykh A, Mora GG, Lanpher BC, Iyer RK, et al. Expanding the phenotypic spectrum in EP300-related Rubinstein-Taybi syndrome. Am J Med Genet. 2015 Feb 25;167(5): 1111–6.	Case: *...The patient..."	*...a relatively broad thumb and great toe,..."	Broad thumb
				*...a relatively broad thumb and great toe,..."	Broad toe
				*...ear dysplasia with "C-shaped" helices..."	Ear anomaly
				*...hirsutism..."	Hirsutism
				*...microcephaly..."	Microcephaly
				*...prominent nose..."	Prominent nose
				*...occipital prominence..."	Prominent occiput
				*...absence of the left kidney..."	Renal anomaly
				*...length of 72.5 cm (<3rd centile..."	Short stature
				*...upslanted palpebral fissures..."	Upward slanted palpebral fissure

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
Russell-Silver	A	Searle C, Johnson D. Russel-Silver syndrome: A historical note and comment on an older adult. Am J Med Genet. 2015 Nov 3;170(2):466–70.	Case: "...This gentleman..."	...fifth finger clinodactyly...	Clinodactyly
				...comparatively large teeth...	Dental anomaly
				...down drawn angle of the mouth...	Downturning corners of mouth
				...high bossed forehead...	Frontal bossing
				...occipital frontal circumference of 54cm (2nd centile)...	Microcephaly
				...Micrognathia was also pronounced...	Micrognathia
				...substantial pes cavus...	Pes cavus
				...weighing 2,100 g (-2.95 SD)...	Prenatal onset growth deficiency
				...Initial failure to thrive...	Short stature
				...triangular shaped face,...	Triangular facies
	B	Perkins RM, Hoang-Xuan MTA. The Russell-Silver syndrome: a case report and brief review of the literature. Pediatr Dermatol. 2002 Nov;19(6):546–9.	Case: "...A 13-month-old boy..."	"...blue sclera..."	Blue sclerae
				"...hypopigmented and hyperpigmented patches on his body..."	Cafe-au-lait spot
				"...Clinodactyly of the fifth digit of the hands bilaterally..."	Clinodactyly
				"...down-turned lips..."	Downturning corners of mouth
				"...hypertelorism..."	Hypertelorism
				"...low-set ears..."	Low-set ear
				"...gross motor delay..."	Mental deficiency
				"...micrognathia..."	Micrognathia
Seckel	A	Shanske A, Caride DG, Menasse-Palmer L, Bogdanow A, Marion RW. Central nervous system anomalies in Seckel syndrome: report of a new family and review of the literature. American Journal of Medical Genetics. 1997 May 16;70(2):155–8.	Case: "...Patient 2..."	"...a large, beaked nose..."	Beaked nose
				"...prominent ears (Fig. 4)..."	Ear anomaly
				"...mild hypospadias..."	Hypospadias
				"...marked delay in achieving psychomotor maturation...."	Mental deficiency
				Table 1: "...microcephaly..."	Microcephaly
				"...prominent eyes..."	Prominent eye
				"...he was small and resembled his sister closely...."	Short stature
				"...simian creases..."	Simian crease
				"...a sloping forehead..."	Sloping forehead
	B		Case: "...Patient 1..."	"...agenesis of corpus callosum..."	Agenesis of corpus callosum
				"...a prominent, beaked nose..."	Beaked nose
				"...mild hypoplasia of the cerebellar vermis...."	Cerebellum hypoplasia
				"...bilateral branched simian creases, clinodactyly of fifth fingers,..."	Clinodactyly
				"...and large, prominent ears with small lobules (Fig. 2)...."	Ear anomaly
				"...She was an active and alert infant without spontaneous or social smile...."	Mental deficiency
				"...microcephaly..."	Microcephaly
				"...prominent eyes..."	Prominent eye
				"...bilateral branched simian creases, clinodactyly of fifth fingers,..."	Simian crease
				"...sloping forehead..."	Sloping forehead
Shprintzen-Goldberg	A	Greally MT, Carey JC, Milewicz DM, Hudgins L, Goldberg RB, Shprintzen RJ, et al. Shprintzen-Goldberg syndrome: a clinical analysis. Am J Med Genet. 1998 Mar 19;76(3):202–12.	Case: "...Shprintzen and Goldberg [1982]..."	Table 1: "...Arachnodactyly..."	Arachnodactyly
				Table 1: "...Camptodactyly..."	Camptodactyly
				Table 1: "...Dolichocephaly..."	Dolichocephaly
				Table 1: "...Downslanting palpebral fissures..."	Downward slanted palpebral fissure
				Table 1: "...Ears, posteriorly angulated..."	Ear anomaly
				Table 1: "...Hypertelorism..."	Hypertelorism
				Table 1: "...Maxillary hypoplasia..."	Micrognathia
				Table 1: "...Pectus carinatum..."	Pectus carinatum
				Table 1: "...Ptosis..."	Ptosis of eyelid
				Table 1: "...Strabismus..."	Strabismus
	B		Case: "...Saal et al. [1995]..."	Table 1: "...Arachnodactyly..."	Arachnodactyly
				Table 1: "...Camptodactyly..."	Camptodactyly
				Table 1: "...Choanal stenosis/ atresia..."	Choanal atresia
				Table 1: "...Craniosynostosis..."	Craniosynostosis
				Table 1: "...Ears, posteriorly angulated..."	Ear anomaly
				Table 1: "...Hypertelorism..."	Hypertelorism
				Table 1: "...Developmental delay..."	Mental deficiency
				Table 1: "...Maxillary hypoplasia..."	Micrognathia
				Table 1: "...Pectus carinatum..."	Pectus carinatum
				Table 1: "...Strabismus..."	Strabismus
Simpson-Golabi-Behmel	A	Halayem S, Hamza M, Maazoul F, Ben Turkia H, Touati M, Tebib N, et al. Distinctive findings in a boy with Simpson-Golabi-Behmel syndrome. Am J Med Genet. 2015 Dec 22;170(4):1035–9.	Case: "...The propositus..."	Table 1: "...Coarse face..."	Coarse facies
				...hepatosple-nomegaly...	Hepatomegaly
				Table 1: "...Hypertelorism..."	Hypertelorism
				...Thoracolumbar kyphosis...	Kyphosis
				Table 1: "...Macrocephaly..."	Macrocephaly
				Table 1: "...Polyhydramniosis..."	Polyhydramnios
				...atypical absence seizures...	Seizure
				...hepatosple-nomegaly...	Splenomegaly
				...accelerated growth (weight and height>97th centile)...	Tall stature
				...umbilical hernia...	Umbilical hernia
	B	Jedraszak G, Girard M, Mellos A, Djeddi D-D, Chardot C, Vanrenterghem A, et al. A patient with Simpson-Golabi-Behmel syndrome, biliary cirrhosis and successful liver transplantation. Am J Med Genet. 2013 Dec 19;164(3):774–7.	Case: "...The boy..."	"...biliary atresia..."	Biliary atresia
				"...diffuse micro- nodular cirrhosis was observed...."	Cirrhosis
				"...facial dysmorphism with coarse features..."	Coarse facies
				"...a left cryptorchidism were noted..."	Cryptorchidism

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
				"...hypertelorism..."	Hypertelorism
				"...occipitofrontal circumference (OFC) 37 cm (>97th centile)..."	Macrocephaly
				"...he was delayed in gross motor functions in a context of axial hypotonia with overgrowth...."	Mental deficiency
				"...polyhydramnios..."	Polyhydramnios
				"...abdominal supernumerary nipples..."	Supernumerary nipple
				"...At the age of 15 months, height, weight and OFC were still above the 97th centile...."	Tall stature
Smith-Fineman-Myers	A	Guion-Almeida ML, Tabith A, Kokitsu-Nakata NM, Zechi RM. Smith-Fineman-Myers syndrome in apparently monozygotic twins. Am J Med Genet. 1998 Sep 23;79(3):205–8.	Case: "...(a) Smith et al. [1980]: 2..."	Table 1: "...Cortical atrophy..."	Cerebral atrophy
				Table 1: "...Dolichocephaly..."	Dolichocephaly
				Table 1: "...Later hypertonia..."	Hypertony
				Table 1: "...microcephaly..."	Microcephaly
				Table 1: "...Micrognathia..."	Micrognathia
				Table 1: "...Small for gestacional age..."	Prenatal onset growth deficiency
				Table 1: "...Prominent upper central incisors..."	Prominent incisor
				Table 1: "...Palpebral ptosis..."	Ptosis of eyelid
				Table 1: "...Seizures..."	Seizure
				Table 1: "...Short stature..."	Short stature
	B		Case: "...(d) Wei et al. [1993]: III-4..."	Table 1: "...Short fingers..."	Brachydactyly
				Table 1: "...Dolichocephaly..."	Dolichocephaly
				Table 1: "...Ocular hypertelorism..."	Hypertelorism
				Table 1: "...Later hypertonia..."	Hypertony
				Table 1: "...Macrostomia..."	Macrostomia
				Table 1: "...Psychomotor retardation..."	Mental deficiency
				Table 1: "...microcephaly..."	Microcephaly
				Table 1: "...Nails anomalies..."	Nail dysplasia
				Table 1: "...Seizures..."	Seizure
				Table 1: "...Short stature..."	Short stature
Spondyloepiphyseal dysplasia congenita	A	Rajab A, Kunze J, Mundlos S. Spondyloepiphyseal dysplasia omani type: A new recessive type of SED with progressive spinal involvement. Am J Med Genet. 2004;126A(4):413–9.	Case: "...A female patient age 4 years..."	"...camptodactyly..."	Camptodactyly
				"...The teeth were small and dentition was delayed with no permanent molars..."	Dental anomaly
				"...limited extension of the elbow and hip joints..."	Elbow limitation
				"...limited extension of the elbow and hip joints..."	Hip limitation
				"...height, however, was 3 cm below the 3rd centile and crown-rump length was 8 cm below the mean..."	Short stature
				"...limitations of movements in the wrist, the elbow, and the hip joints were observed..."	Wrist limitation
	B	Kawano O, Nakamura A, Morikawa S, Uetake K, Ishizu K, Tajima T. Spondyloepiphyseal dysplasia congenita caused by double heterozygous mutations in COL2A1. Am J Med Genet. 2015 Apr 21;167(7):1578–81.	Case: "...The patient..."	...myopia...	Myopia
				...Mild odontoid hypoplasia...	Odontoid hypoplasia
				...Coxa vara and short femoral necks were noted (Fig. 1C)...	Short femoral neck
				...Short limbs were noted during pregnancy...	Short limb
				...birth length was 42.6 cm (-3.5 SD)...	Short stature
				...spine was characterized by platyspondyly with dorsal vertebral flattening...	Vertebral anomaly
Toriello-carey	A	Chinen Y, Tohma T, Izumikawa Y, Taketomi H, Iha T, Ohta T, et al. Two sisters with Toriello-Carey syndrome. American Journal of Medical Genetics. 1999 Nov 26;87(3):262–4.	Case: "...Patient 1..."	Table 1: "...Cerebellum hypoplasia..."	Cerebellum hypoplasia
				"...hypoplastic cerebellar hemisphere, vermis, and corpus callosum (Fig. 1c)...."	Cerebral atrophy
				"...Severe tetralogy of Fallot consisted of ventricular septal defect,..."	Fallot
				"...telecanthus..."	Hypertelorism
				"...lax finger joints..."	Joint hypermobility
				"...OFC 45.2 cm (−2.9 SD)..."	Microcephaly
				"...Her weight was 11.5 kg (−2.3 SD), height 89.5 cm (−3.1 SD),..."	Short stature
				"...a small umbilical hernia..."	Umbilical hernia
				"...cleft uvula..."	Uvula bifid
				"...showed coronal notching of lumbar vertebral bodies..."	Vertebral anomaly
	B		Case: "...Patient 2..."	"...agenesis of the corpus callosum (Fig. 2c)...."	Agenesis of corpus callosum
				"...anteverted nostrils..."	Anteverted nare
				"...hypoplastic cerebellar hemisphere..."	Cerebellum hypoplasia
				"...aplastic cerebellar vermis..."	Cerebral atrophy
				"...cleft palate..."	Cleft palate
				"...Neonatal cyanosis was caused by severe tetralogy of Fallot..."	Fallot
				"...and dyspnea caused by glossoptosis..."	Glossoptosis
				"...microretrognathia..."	Micrognathia
				"...Hypoplasia of the left optic disc was suggested by an ophthalmologist..."	Optic nerve hypoplasia
				"...patent ductus arteriosus..."	Patent ductus arteriosus
Williams	A	Castorina P, Selicorni A, Bedeschi F, Dalprà L, Larizza L. Genotype-phenotype correlation in two sets of monozygotic twins with Williams syndrome. American Journal of Medical Genetics. Wiley Subscription Services, Inc., A Wiley Company; 1997 Mar 3;69(1):107–11.	Case: "...Family 2: Twin B..."	Table 1: "...Clinodactyly of fifth finger..."	Clinodactyly
				Table 1: "..."Coarse" face..."	Coarse facies
				Table 1: "...Sparse eyelashes..."	Eyelash hypoplasia
				Table 1: "...Facial asymmetry..."	Facial asymmetry
				Table 1: "...Long philtrum..."	Long philtrum
				Table 1: "...Large mouth..."	Macrostomia
				Table 1: "...Mental retardation..."	Mental deficiency
				Table 1: "...microcephaly..."	Microcephaly
				Table 1: "...SVAS..."	Supravalvular aortic stenosis

Syndrome	Groupe	Article source	Case report	Signe / symptôme	Equivalent SYNDROC
	B		Case: "...Family 2: Twin A..."	Table 1: "...Clinodactyly of fifth finger..."	Clinodactyly
				Table 1: "..."Coarse" face..."	Coarse facies
				Table 1: "...Epichantal folds..."	Epicanthus
				Table 1: "...Sparse eyelashes..."	Eyelash hypoplasia
				Table 1: "...Long philtrum..."	Long philtrum
				Table 1: "...Large mouth..."	Macrostomia
				Table 1: "...Mental retardation..."	Mental deficiency
				Table 1: "...microcephaly..."	Microcephaly
				Table 1: "...SVAS..."	Supravalvular aortic stenosis