



# 3D CT digital reconstruction and analysis of Gorlin and Goltz syndrome

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## OBJECTIVES

In 1960 Gorlin and Goltz defined a syndrome with a principal triad of multiple basal cell nevi, jaw odontogenic keratocysts and skeletal anomalies. This syndrome has been given several alternative names, but it is mainly known as “basal cell nevus syndrome” (BCNS). Other clinical characteristics include macrocephaly, ectopic calcifications, plantar and palmar pits, central nervous system and ocular lesions, and fairly typical facial features with frontal bossing and hypertelorism. The estimated prevalence of BCNS ranges from 1:57,000 to 1:256,000 and affects both genders equally. The syndrome is inherited in an autosomal dominant manner and is with a strong pathogenic variant penetrance. According to recent molecular genetic studies, approximately 35–50% of cases result from de novo germline mutations. It is estimated that 30–50% of patients suffering from BCNS are not aware of any other affected family member.

## MATERIAL AND METHODS

The present patients case reposts demonstrate the presence of all typical features such as multiple cysts in the facial skeleton, asymmetry of the face in the 3D images, skin lesions like a pigmented nevi, basaliomas, cutaneous keratocysts and macrocephaly, tumors etc. The dentists, orthodontists, radiologists or neurologists are the first to encounter this syndrome. Diagnosis is based on a panoramic image, most of which is complemented by 3D CT digital reconstruction and analysis. It is followed by neurological, dermatological and histological examination. The basis, however, is clinical and molecular genetics of rare disease. Treatment is surgical and consists in extirpation of the cyst, without bone augmentation, especially in pediatric patients. Case report includes a retrospective analysis of a representative Czech cohort with BCNS from within the last decade. The aim of study is to analyze of 3D reconstruction options during rare diseases diagnoses and therapy.

## CASE REPORTS

### Case report 1

22 year old man, in a history basal and pigment nevi on the neck and abdomen, in 3/2013 first examination, 4/2013 surgical therapy (removal of cysts loco 18,28,38,48), in this time without evidence of cyst in the facial skeleton

OPG –cysts loco 18,28,38,48



OPG –post oper.



### Case report 2

20 year old woman, in a history in 1 year excision of the upper lip lesion (dermoid), pollen allergy, in 5/2009 first examination, 6/2009 – surgical treatment (removal of cyst I.23), 8/2010 – removal of cyst loco 43, 12/2011 – removal of cyst I.38, 4/2014 – removal cyst of I.28

OPG – cyst loco 23



OPG - post oper.



### Case report 3

15 year old girl, in a history medulloblastoma with hydrocephalus, onkology therapy, basaliomas, hypopituitarismus, allergy Augmentin, in 7/2013 surgical therapy (removal of multiple cyst of maxilla and mandibula), in 12/2014 – removal of cyst loco 33

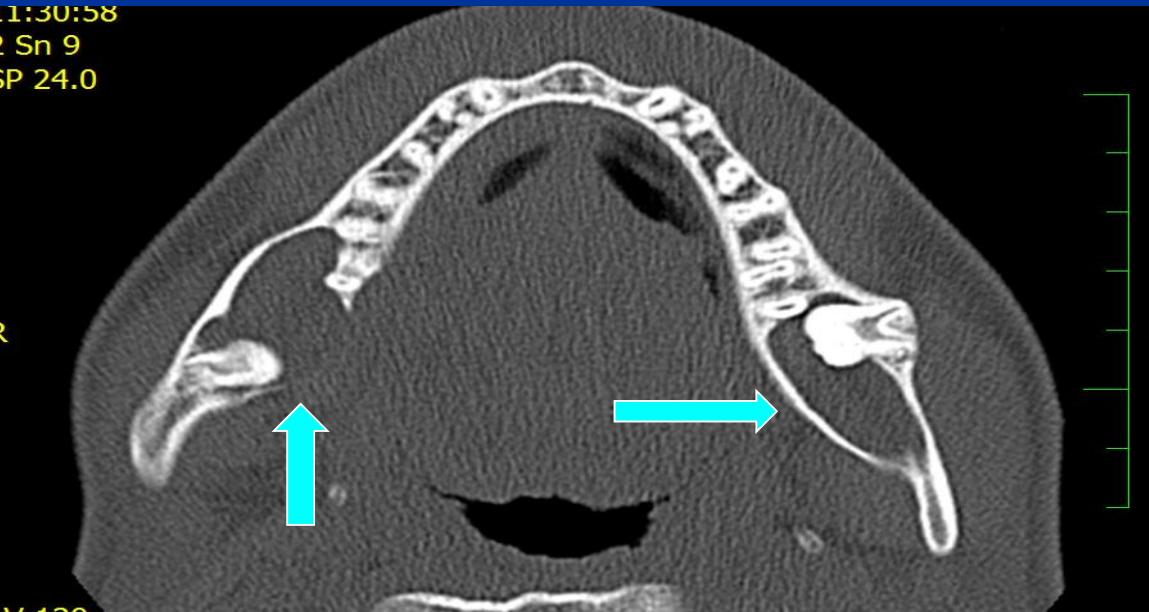
OPG – multiple cyst of maxila and mandibula



OPG – cyst of mandibula loco 33



CT – cysts loco 38,48



CT – post oper.



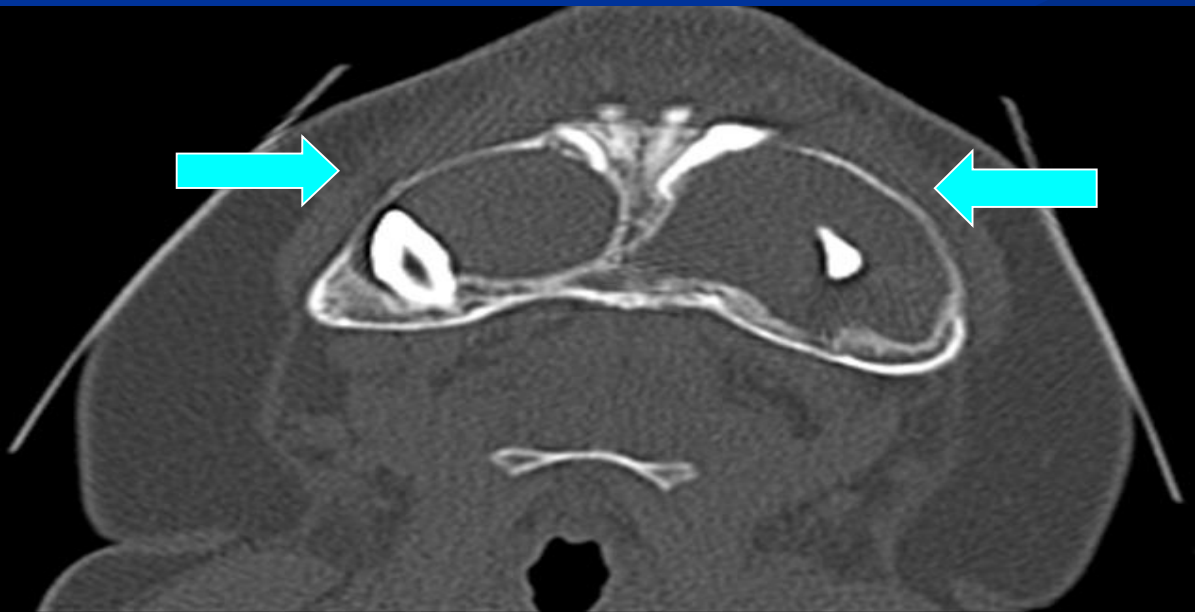
CT – cyst loco 23 (coronal view)



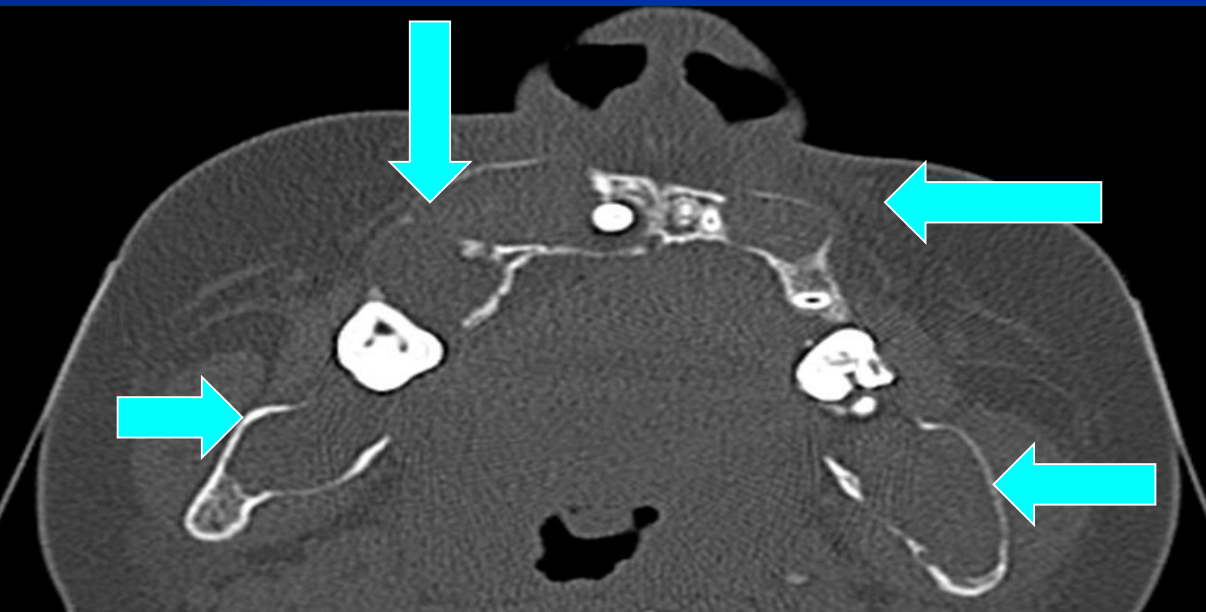
CT – cyst loco 23 (sagittal view)



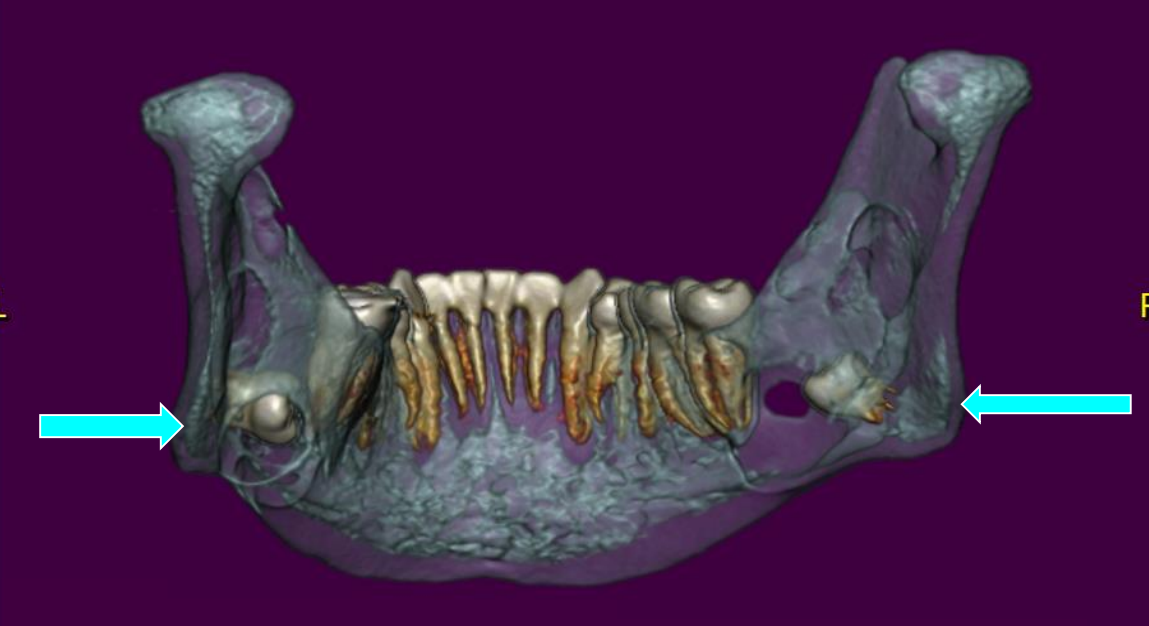
CT – two cyst of mandibula



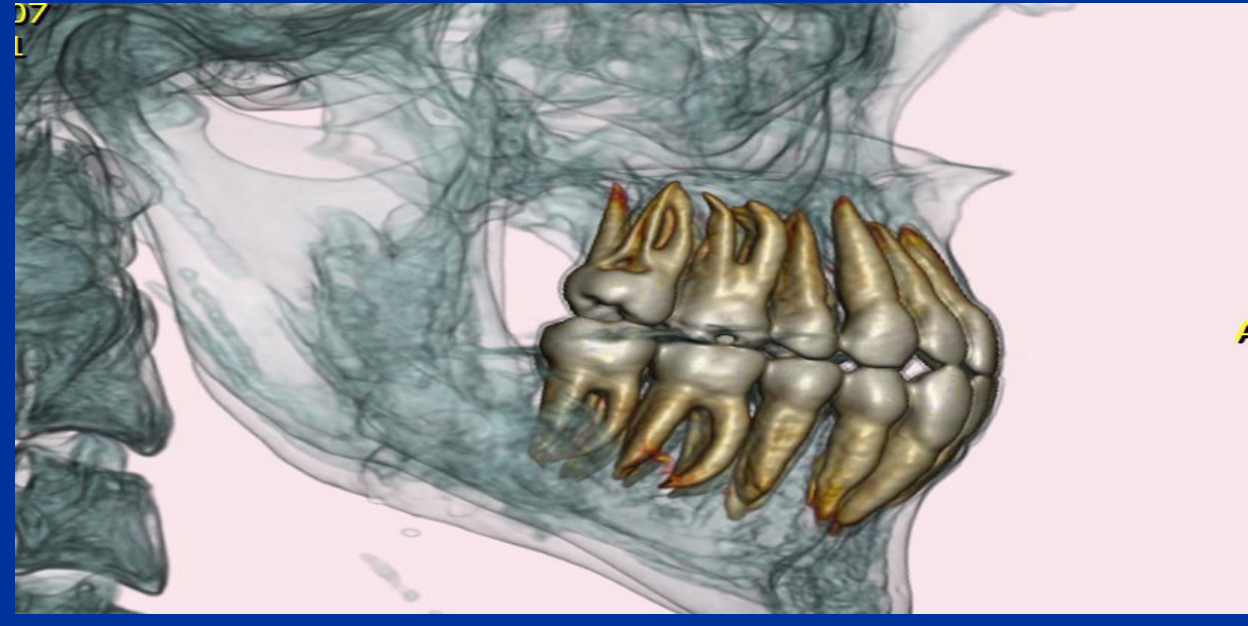
CT – multiple cysts of maxila



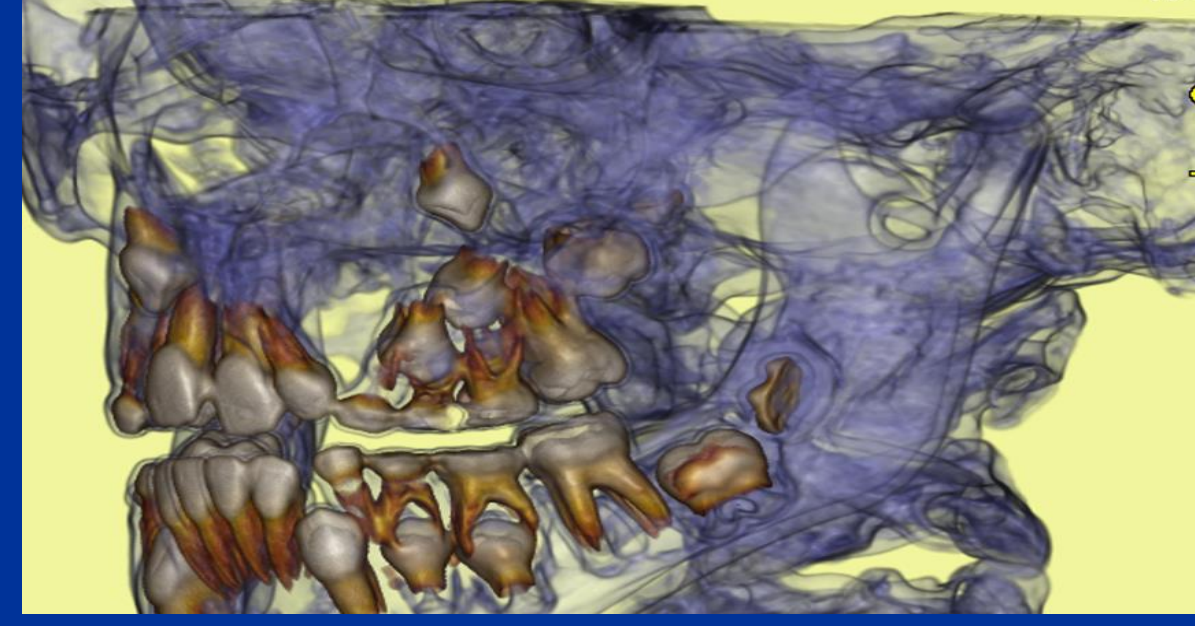
3D CT reconstruction



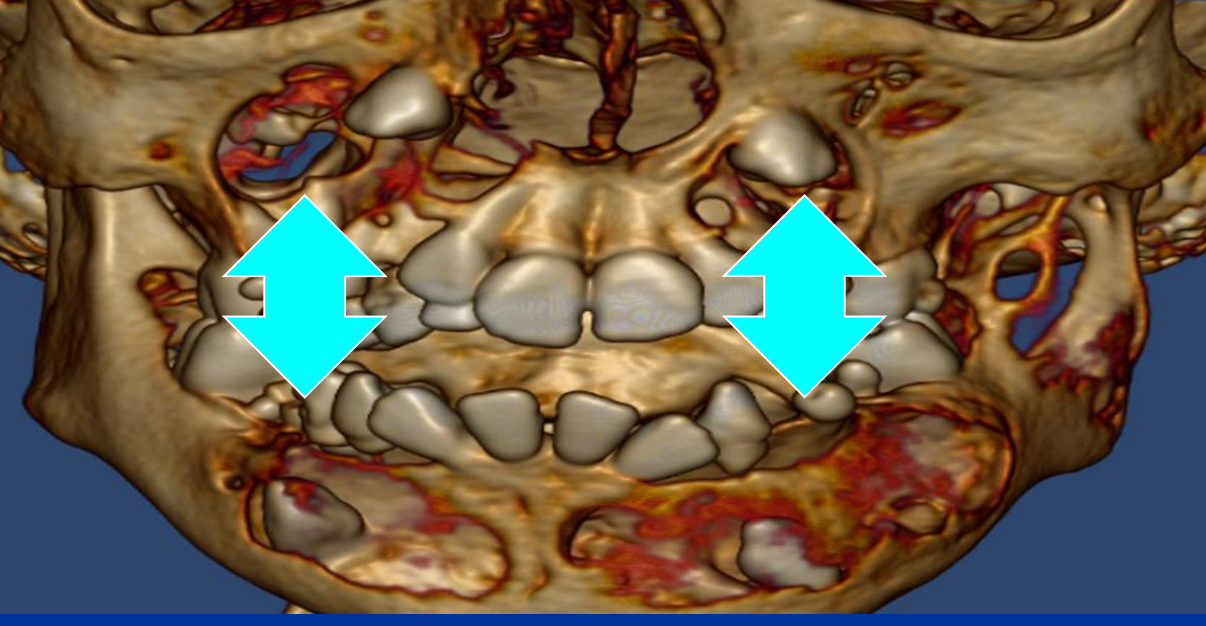
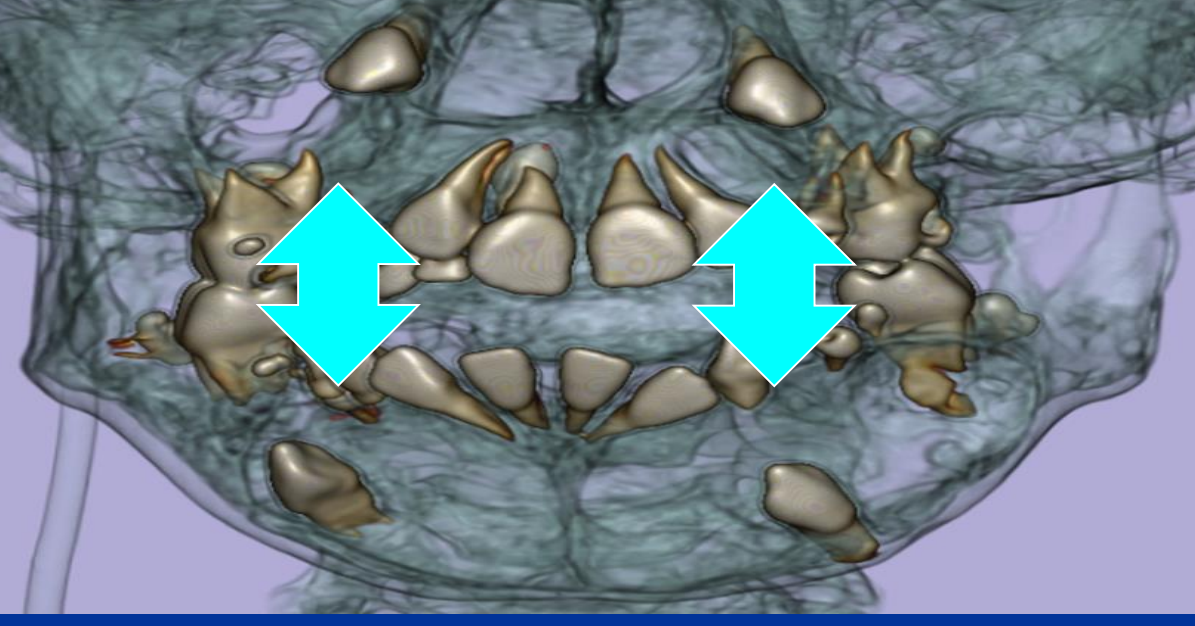
3D CT reconstruction – post oper.



3D CT reconstruction – cyst loco 23



3D CT reconstruction – multiple cysts of maxila aned mandibula



## DISCUSSION

Multislice computed tomography (MSCT) is new 3D radiological techniques for imaging of facial skeleton. Conventional multislice computed tomography due to radiation dose is used mainly for forensic identification. The MSCT device Aquilion 64 (Toshiba Medical Systems Europe BV, Zoetermeer 2718RP The Netherlands) was used in our study. From the raw data were generated the volume datasets, which were focused on the dental area. 3D images of bones were creating using VRT (volume rendering technique), basic visualization techniques mainly cutting, thresholding) and manual segmentation. The average size of isotropic voxel was 0.4 mm.

## CONCLUSION

Awareness of BCNS among medical specialists who may encounter these patients in the first place (e.g. orthodontists, dentists, radiologists, neurologists) is important for its early diagnosis. Patients reporting lower jaw paresthesia should not be disregarded. The long-term consequences of the BCNS are associated with server morbidity and markedly decreased quality of life in patients. 3D reconstruction of skull helps us precise plan diagnoses and treatment. In this regard, early diagnosis represents an opportunity for effective management of this severe and progressive rare disorder.

### Clinical Characteristics of Patients with Gorlin Goltz Syndrome

	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5	Presented case (Patient 6)	Patient 7
Pathogenic Variant	c.2703+1G>C	c.2726delAlmsTGC (p.Asp909Valfs*16)	c.838delG (p.Glu280Lysfs*3)	c.2309-2311delGAGinsA (p.Arg770Hisfs*19)	c.2309-2311delGAGinsA (p.Arg770Hisfs*19)	c.2929dupT (p.Tyr977Leufs*16)	c.3181dupG (p.Ala1061Glyfs*84)
Sex	F	F	M	M	F	M	M
Age of evaluation	15y	9y	16y	13m	prenatally	15y	7y
Parental analysis (mother/father)	-/-	-/-	na	+/- (+ maternal grandfather and uncle)	+/- (+ maternal grandfather and uncle)	-/-	-/-
Macrocephaly	+	+	+	+	+	+	+
Jaw odontogenic keratocyst	+	+	+	+	+	+	+
Skin lesions	trichofolliculoma of upper lip, pigmented nevi, cutaneous keratocyst	basaliomas, pigmented nevi	basaliomas, pigmented nevi	basaliomas, pigmented nevi	pigmented nevi	basaliomas, pigmented nevi	pigmented nevi, nevus flammeus
Tumors	-	medulloblastoma	-	-	fibroma of left ventricle	-	-
Others	-	severe kyphoscoliosis and lordosis, cysts of eyelids, bone cysts in vertebrae	right kidney cyst, mitral and aortal regurgitation	pancreatic cyst, brain venous angioma, brain arachnoid cyst	strabism, posthypoxic encephalopathy after CPR, intellectual disability	VSD, mitral valve prolapse, arrhythmia, kyphoscoliosis	kyphosis, ribs and vertebral malformation, brain arachnoid cyst, hypermetropia
Treatment	extirpation of odontogenic keratocysts and excision of trichofolliculoma	extirpation of odontogenic keratocysts, excision of basaliomas, resection and chemotherapy of medulloblastoma	extirpation of odontogenic keratocysts, excision of basaliomas	extirpation of odontogenic keratocysts, excision of basaliomas	extirpation of odontogenic keratocysts, implantation of defibrillator	extirpation of odontogenic keratocysts, excision of basaliomas	extirpation of odontogenic keratocysts



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