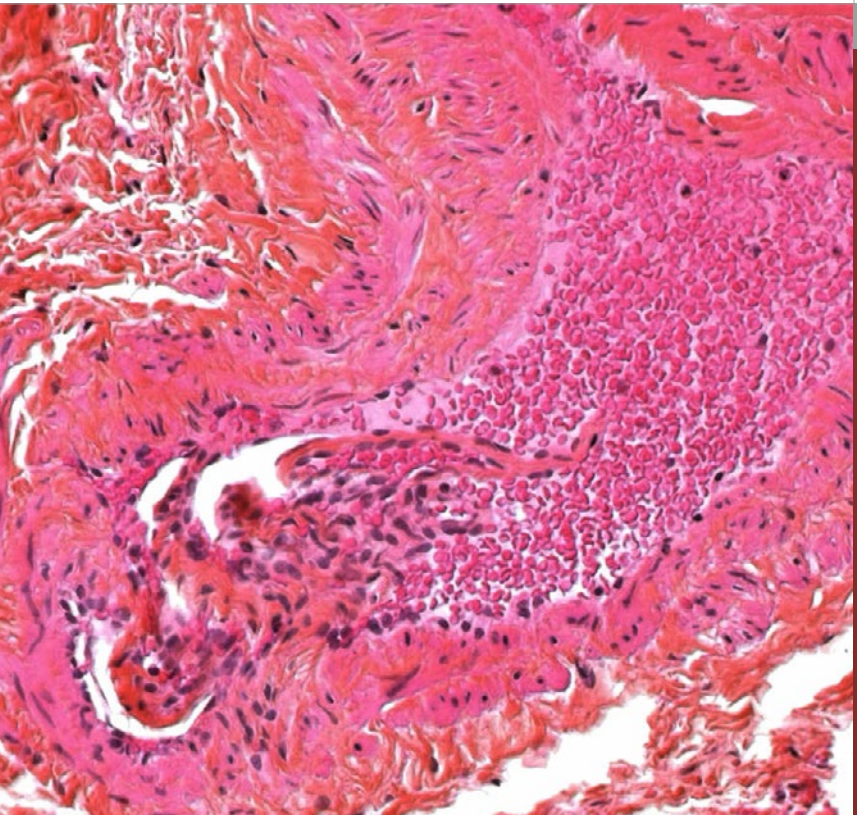
SWISS SOCIETY OF NEONATOLOGY

A congenital finding with a rich presentation



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Title figure:
Hemangioma (source: www.humpath.com)

Congenital hemangiomas (CH) are rare childhood vascular tumors. In contrast to the very common infantile hemangiomas (IH), they are fully developed at birth. They usually present as skin-colored or bluish lumps located on the head, neck or limbs and, in rare instances, internal organs such as the liver may also be affected (1). Two major subtypes have been recognized based on their natural history: a) rapidly involuting congenital hemangioma (RICH) and b) non-involuting congenital hemangioma (NICH).

We present two newborns with CH, highlight the initial management and illustrate the further course of their vascular tumors.

CASE REPORT 1

This male infant was born by spontaneous vaginal delivery at 41 2/7 weeks of gestation to a 34-year-old G3/P2 after an uneventful pregnancy. The infant adapted well with Apgar scores of 6, 9 and 10 at 1, 5 and 10 minutes, respectively. Birth weight was 4100 g. Immediately after delivery, a solid, reddish to violaceous mass with an anemic halo measuring 7.5 cm × 6 cm × 4 cm was noted on the right lower leg (Fig. 1). Physical examination was otherwise normal. The neonate was first observed in the maternity clinic, where an ultrasound of this mass was performed but could not exclude a malignant tumor. Therefore, the neonate was transported to the University Children's Hospital of Zurich on day one of life for further work-up and management. On arrival, he was well-appearing without any cardiorespiratory compromise.

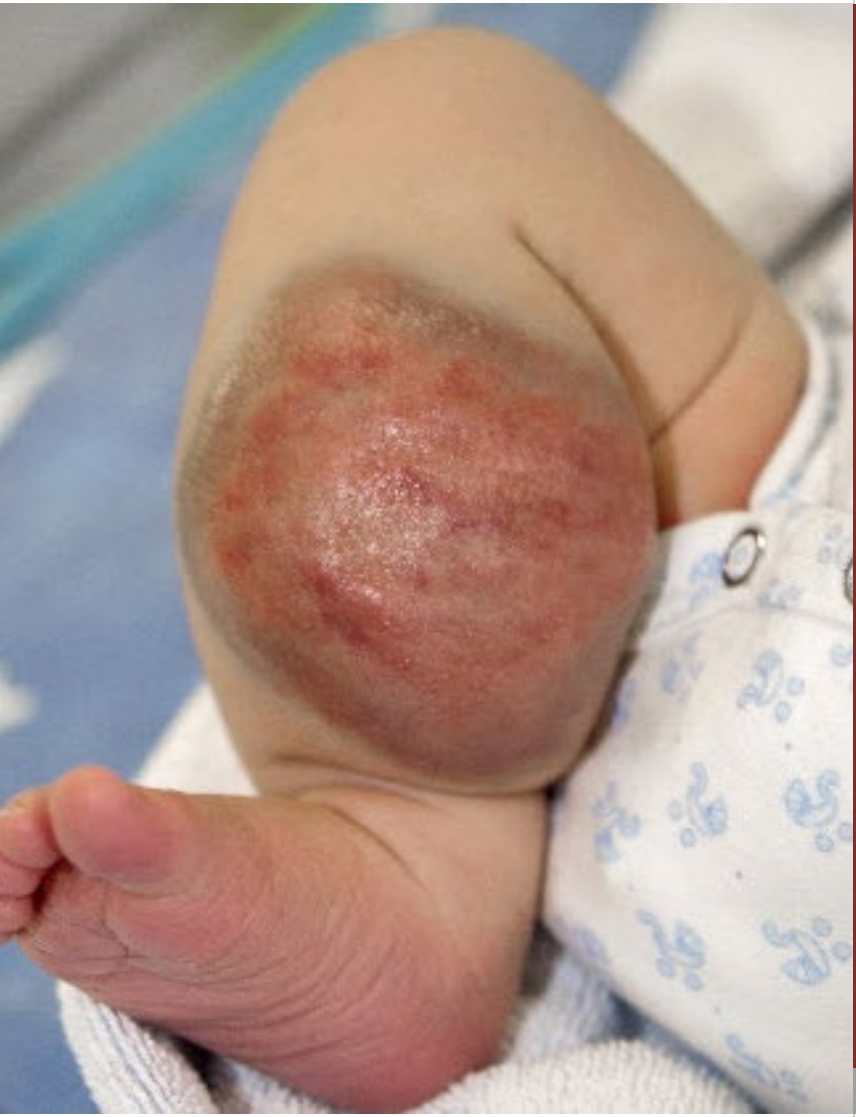

Fig. 1

Patient 1 (DOL 1): solid, violaceous mass with a halo on the right lower leg.

Based on the clinical presentation, RICH was suspected. Findings of Doppler ultrasound were compatible with this diagnosis (Fig. 2). A complete blood count as well as coagulation studies including d-dimers were all within normal limits. An ultrasound of the abdomen was normal as well, excluding an involvement of internal organs.

The boy was discharged home on the fourth day of life. As expected for RICH, the lesion was rapidly regressive within the first few months of life (Fig. 3) and regression was almost complete after 14 months (Fig. 4).

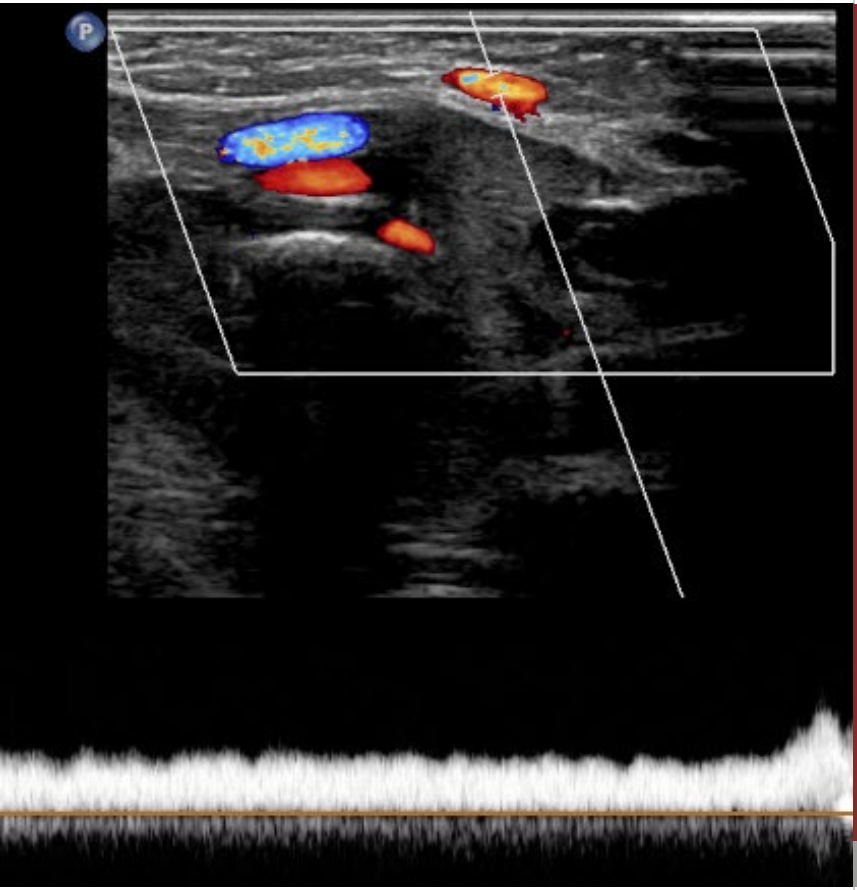

Fig. 2

Patient 1 (DOL 2): Doppler ultrasound examination demonstrating a fast-flow-lesion on the right lower leg.

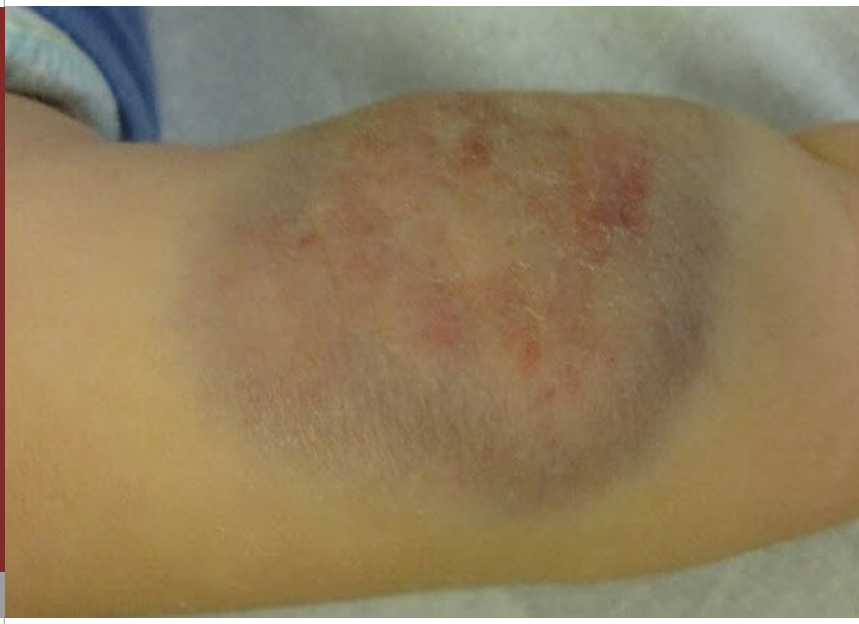

Fig. 3

Patient 1 (age of 2 months): impressive spontaneous regression.

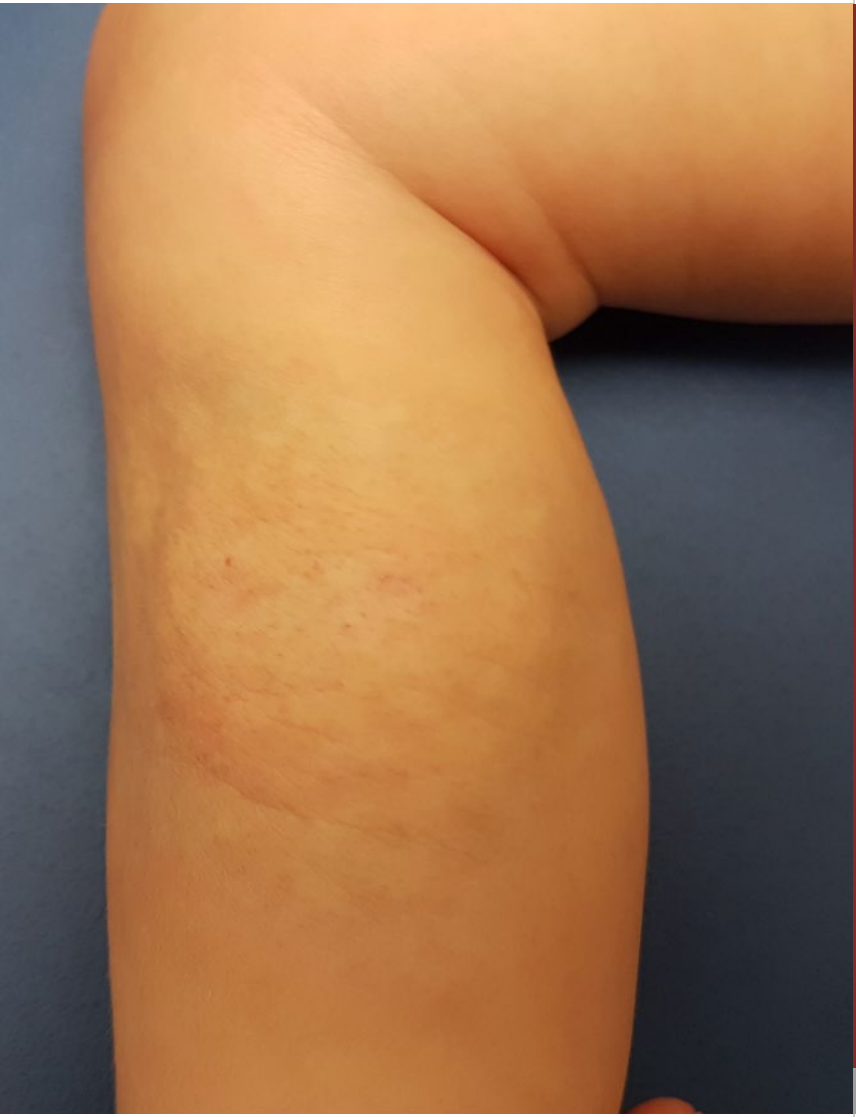

Fig. 4

Patient 1 (age of 14 months): almost complete regression.

CASE REPORT 2

This female infant was born by spontaneous vaginal delivery at 40 1/7 weeks of gestation to a 29-year-old G1/P1 after an uncomplicated pregnancy. She adapted well with Apgar scores of 8, 9 and 10 at 1, 5 and 10 minutes, respectively. Birth weight was 2800 g. After delivery, a giant, bluish, soft mass on the right shoulder was noted (Fig. 5). Apart from this, clinical examination was unremarkable without any signs of cardiorespiratory problems or cardiac failure.

Doppler ultrasound examination showed a subcutaneous, well-demarcated, hyperechogenic mass, measuring 9 cm × 4 cm × 1.5 cm with arterial and venous flow signals. Based on these findings, RICH was suspected. To exclude high-output cardiac failure due to AV-shunting within the lesion, echocardiography was performed and was within normal limits. Blood count and coagulation studies were normal except for a slightly decreased platelet count (minimum 66 G/l on day five).

Because of the impressive size of this tumor and thrombocytopenia the neonate was transported to the University Children's Hospital of Zurich, where an MRI was performed to exclude possible differential diagnoses (Fig. 6). This demonstrated a highly vascularized, well-demarcated subcutaneous lesion, typical of a hemangioma. Due to thrombocytopenia, the child was monitored for the possible development

of a Kasabach-Merritt phenomenon (KMP), but the platelet counts spontaneously increased to normal limits. Because of the typical clinical and imaging findings, as well as spontaneous resolution of thrombocytopenia, no biopsy was performed.

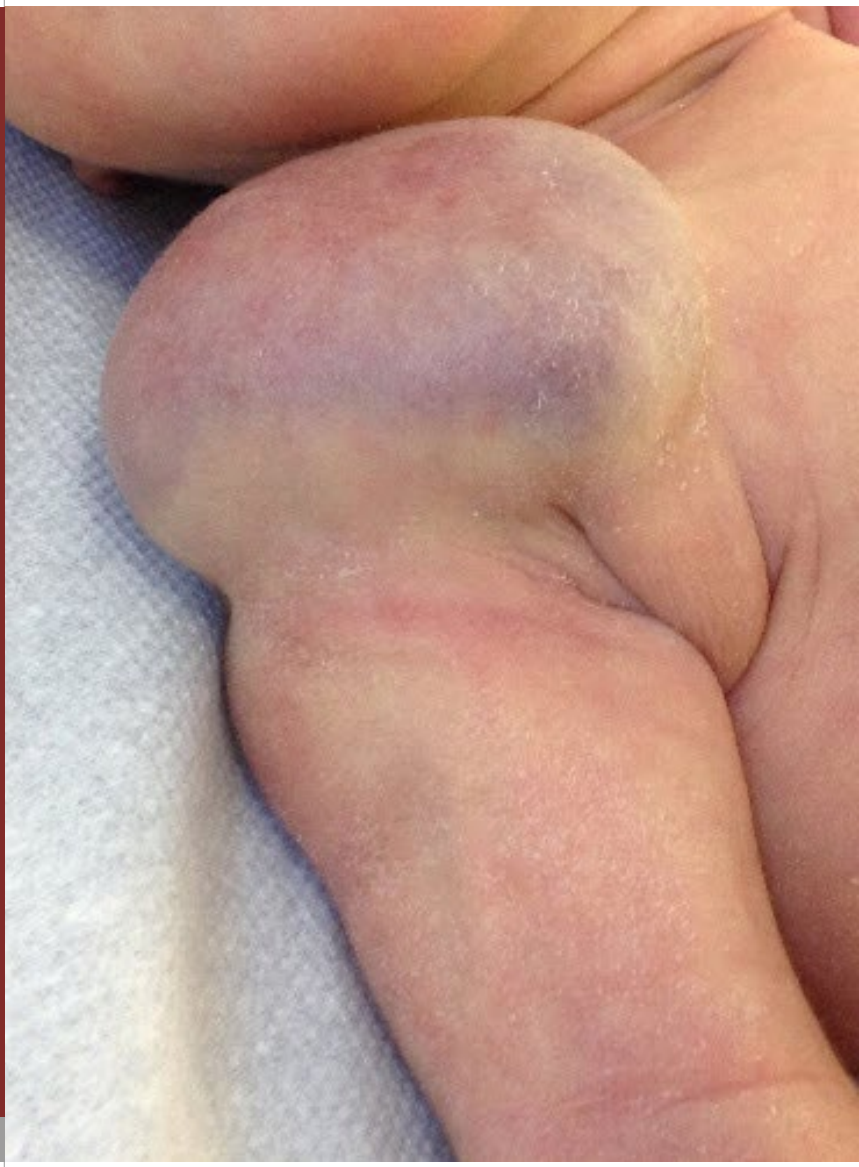

Fig. 5

Patient 2 (DOL 7): bluish soft mass with telangiectasia on the right shoulder.

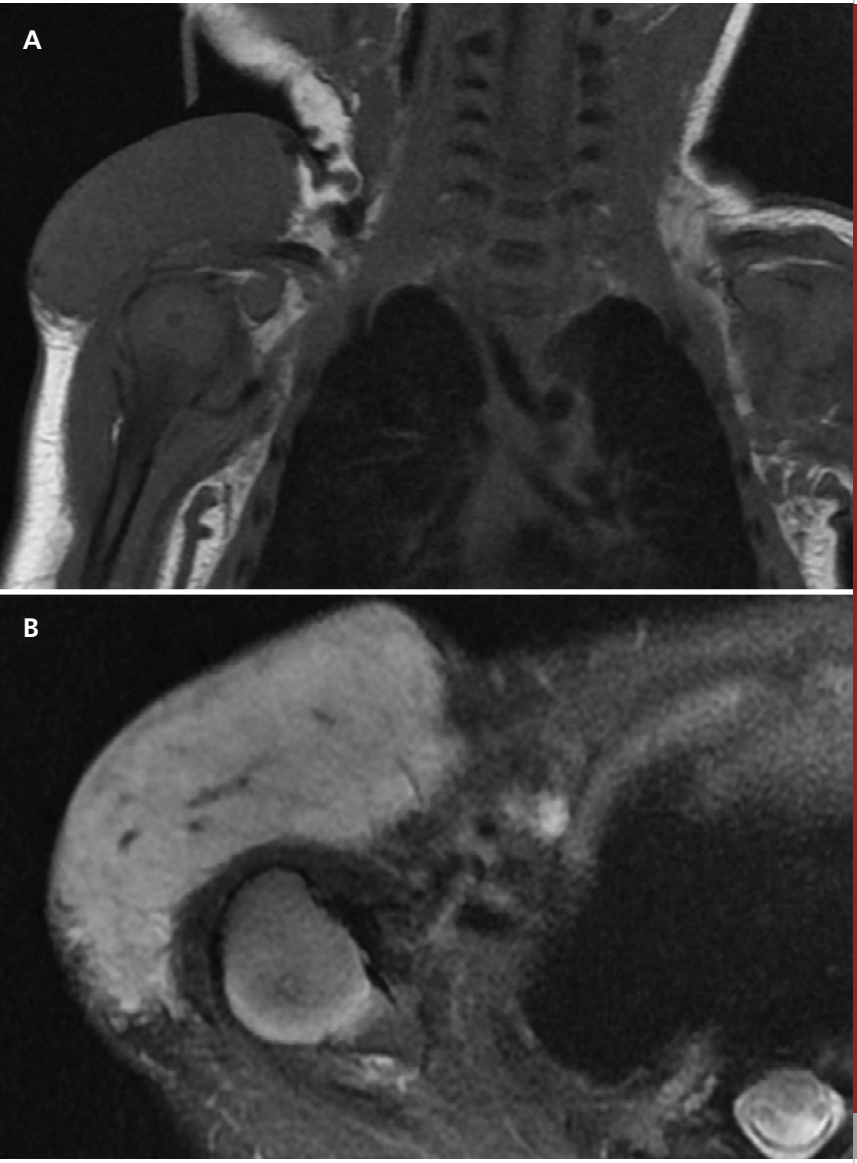

Fig. 6

Patient 2: MRI of the right shoulder (DOL 7) outlining a highly vascularized, well-demarcated subcutaneous lesion, typical of a hemangioma, (A: T1-weighted image; B: T2 weighted image).

The girl was discharged in excellent condition on the eighth day of life. Regular follow-up was scheduled, and, at 4 and especially at 6 months of life, spontaneous regression became obvious (Fig. 7 and 8). At 12 months of life, regression was almost complete resulting in a partially atrophic plaque (Fig. 9).

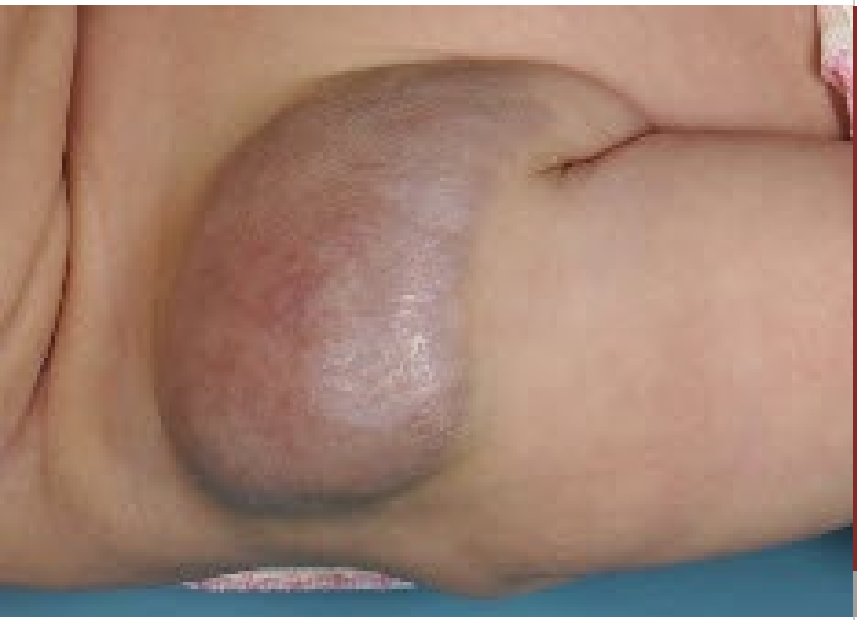

Fig. 7

Patient 2 (age of 4 months): partial spontaneous regression.

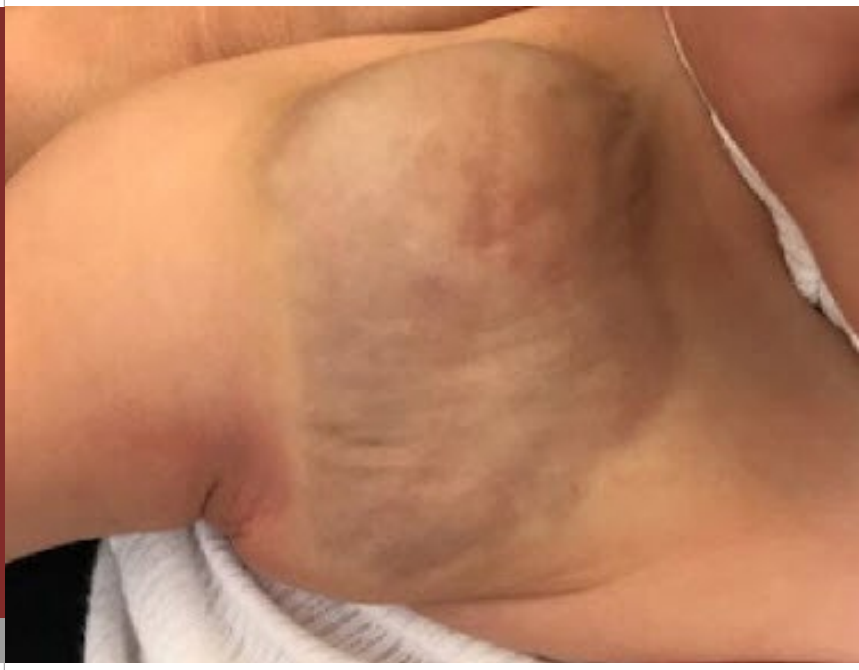

Fig. 8

Patient 2 (age of 8 months): partial spontaneous regression.

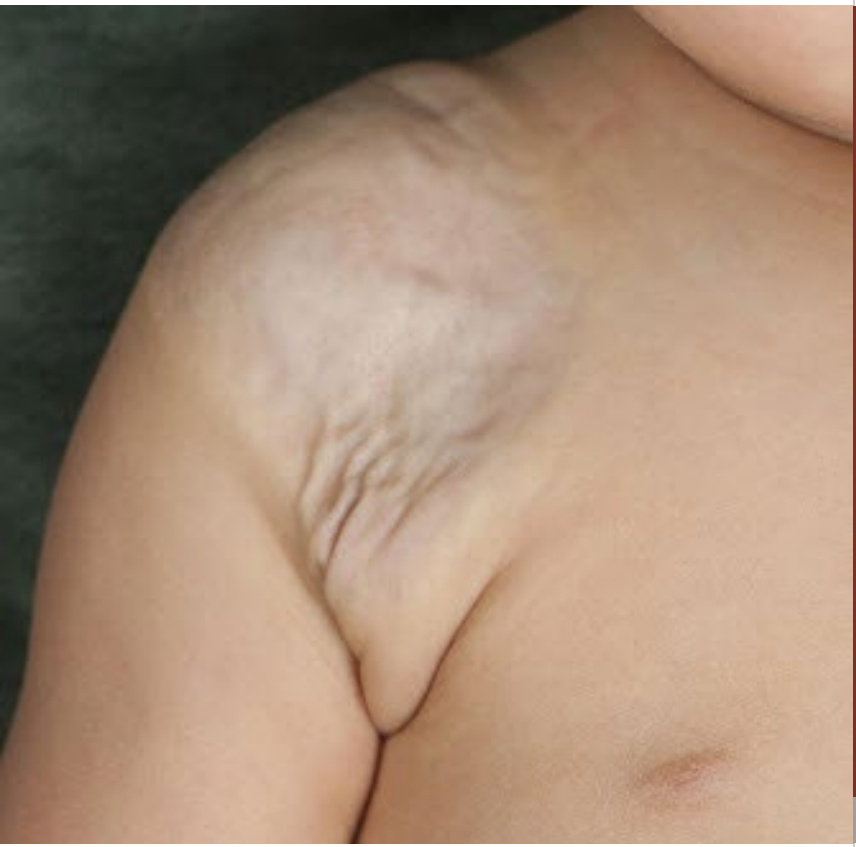

Fig. 9

Patient 2 (age of 12 months): partially atrophic plaque.

DISCUSSION

CH are uncommon vascular tumors fully grown at birth. The precise incidence of CH is not known. In contrast to IH, there is no association with female sex, prematurity or multiple gestation pregnancies. In CH, extracutaneous manifestations are extremely rare (1).

The pathogenesis of CH is poorly understood. However, they seem to be entities clearly distinct from IH. This distinction is based on their very different clinical course, their failure to respond to beta-blocker treatment (2), as well as histological differences. Most importantly, CH stain negatively on immunohistochemistry for GLUT-1 (glucose transporter 1), which is strongly positive in all phases of IH development and regression (1).

CH present as solitary, skin-colored, reddish or violaceous tumors or plaques, often surrounded by a pale halo. Central telangiectasias may also occur, however, they do not have the strawberry presentation of superficial or mixed IH. On palpation, lesions are typically warmer than the surrounding skin. The size may vary from a few to more than 10 cm. CH are divided into those that involute and those that do not. RICH are much more common than NICH. Involution often begins early in the postnatal period (1); the involved mechanisms are unknown.

RICH usually resolve within the first 6 – 14 months of life, often leaving an atrophic area. Even if they are

very large at birth, they can involute completely (1). Involution in utero has also rarely been described. NICH are typically flatter than RICH, do not regress spontaneously but grow in proportion with the child (1). If necessary, they are treated surgically. In rare instances, CH involute rapidly during the first year of life, but fail to regress completely. In these cases, they are called partially involuting congenital hemangioma (PICH) (1).

An important early complication of CH is extensive arteriovenous shunting leading to high-output cardiac failure. This typically occurs in patients with large lesions and, therefore, frequent clinical assessments and evaluation of cardiac function by echocardiography are strongly recommended. Rarely, ulceration with bleeding may occur. In rare cases, bleeding is severe requiring immediate intervention such as embolization or surgical intervention. In addition, transient thrombocytopenia, consumption coagulopathy (usually mild) and anemia can develop, as in our second patient (1, 2). However, true KMP is exceptional in CH and much more typical of other vascular tumors such as tufted angioma or kaposiform hemangioendothelioma.

The differential diagnosis of CH is wide and includes benign vascular tumors, vascular malformations and nonvascular tumors including malignancies (Table 1). Imaging findings can help to clarify the diagnosis.

On Doppler ultrasound examination, IH and CH are persistent fast flow lesions. Complete blood count and coagulation studies help to differentiate CH from vascular tumors frequently presenting with KMP.

One should refrain from assuming that a bluish or reddish discoloration proves the vascular nature of a tumor. We therefore strongly recommend obtaining a biopsy of any congenital bluish mass that does not display the clear-cut features of a CH on ultrasound or that does not behave as expected (especially if growing in size).

Type of lesion	Presentation at birth	Clinical appearance	Coagulation	Ultrasound	Histology
Congenital hemangioma	fully grown at birth	solitary tumor (skin-coloured, reddish, violaceous), pale halo, central teleangiectasias	(thrombocytopenia)	fast-flow lesion	Glut-1 negative
Infantile hemangioma	1–2 (12) weeks after birth	macule of branching capillary blood vessels at birth, rapid proliferation within the first weeks of life		fast-flow lesion	Glut-1 positive
Tufted hemangioma	can be present	solitary or multiple red to violaceous tumors	KMP	fast-flow lesion	typical histology
Kaposiform hemanioendothelioma	can be present	rare subcutaneous tumor, progressive growth	KMP	fast-flow lesion	typical histology
Venous malformation	usually present	soft, compressible mass		slow-flow lesion	
Myofibromatoma	can be present	firm or rubbery mass			typical histology
Lipoblastoma	can be present	well circumscribed soft mass			typical histology
Malignant	can be present	signs of malignancy			

Differential diagnoses of bluish or reddish cutaneous/subcutaneous lesions present at birth (KMP: Kasabach-Merritt phenomenon).

The management of CH must be tailored to the individual patient, depending on the clinical presentation, the size, the location and possible complications. Doppler ultrasound examination should be performed by an experienced pediatric radiologist in any suspected CH, and biopsy is recommended in equivocal cases. Laboratory investigations (complete blood count and coagulation studies including D-dimers) should be performed to exclude other vascular tumors. Usually, treatment is not necessary in RICH, except perhaps corrective surgery after full regression in some patients. For NICH surgical excision may be required. Propranolol is not a therapeutic option in CH.

The differential diagnosis of congenital bluish or reddish masses is wide. In cases with typical ultrasound findings of a hemangioma and in the absence of severe coagulation disturbances, CH may be suspected and followed clinically. As CH may present as large tumors, it is important to guide parents accordingly, providing them with detailed information about the clinical course, possible complications and treatment options for non- or partially involuting lesions as well as residual skin changes after involution.

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