Macropephaly-Cutis Marmorata: Case Report and Review of Literature

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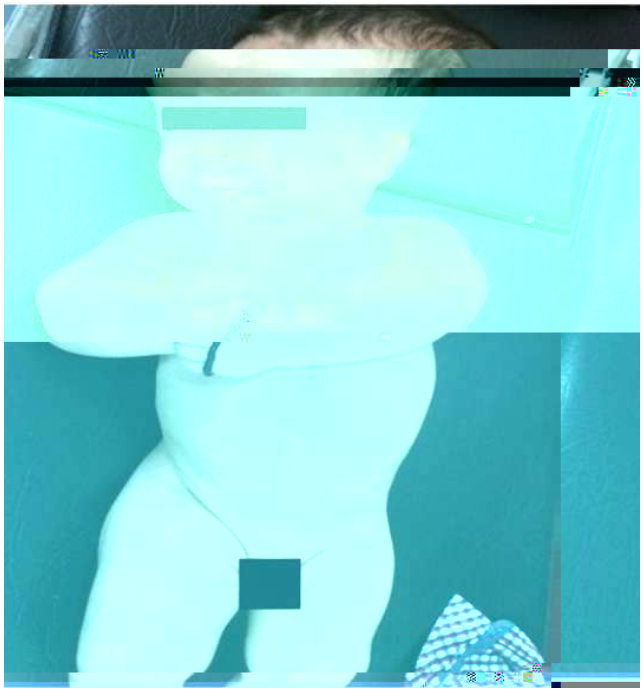

Figure 2: Appearance of cutis marmorata with a so f consistency and an excess of skin folds.

T e MRI showed cortical atrophy. Abdominal echocardiography and the urinary sha f revealed hepatomegaly and nephrenomegaly. T e fundus examination was normal outside of a hyperemia. T ere was had no abnormalities on echocardiography and the biological assessment was normal (blood glucose, renal and hepatic function without abnormalities with chromatography of organic acids in the urine) and the karyotype is in progress. In front of this clinical picture: macrocephaly, vascular anomaly and hemi-hypertrophy of the body. T e diagnosis of macrocephaly-capillary malformation syndrome was retained.

Discussion

Macrocephaly - capillary malformation (M-MC) was f rst described by Moore et al. [1], and Clayton-Smith et al. [2]. In 1997 as macrocépahlie-cutis marmorata telegiectasia congenital syndrome (M-CMTC) respectively, in 9 and 13 children, all patients had cutis marmorata telangiectatica congenita (CMTC), hypotonia and / or psyar%-c ή

Diagnostic Criteria	Wright et al. [9].	Martinez-Glez et al. [10].	Mirzaa et al. [5].
Major	2 criteria required	3 criteria required	
	I- Macrocephaly*	I- Macrocephaly*	I- Early growth (brain> somatic tissues). Progressive megalencephaly*.
	II- Capillary malformation*	II- Capillary malformation*.	II- Capillary malformation: face and medio-corporal*.
		II- Hypertrophy/asymmetry*. IV- Anomaly of neuroimaging: ventruculomegaly, spinal/cerebellar asymmetry, hernia brain tonsils, cavum septum pellucidum or cavum vergae.	III- Abnormalities of the distal limbs. Syndactyly (2-3, 3-4, 2-3-4, toe or finger).
			IV- Cortical brain malformations: Polymicrogyria.
			V- Connective tissue dysplasia*: hyperelasticity of the skin, joint hypermobility and thick, pasty subcutaneous tissue.
Minor	At least 2 criteria	At least 2 criteria	
	1- Medio-facial capillary malformation.	1- Medio-facial capillary malformation.	1- Selective overgrowth of the brain:
	2- Developmental delay*.	2- Developmental delay*.	ventriculomegaly / hydrocephalus.
	3- Neonatal hypotonia*.	3- Neonatal hypotonia*.	Cerebellar ectopia of the tonsils.
	4- Poly or Syndactylia.	4- Syndactyly or polydactyly.	Callous (mega) body abnormally thick.
	5- Frontal bumps.	5- Frontal bumps*.	2- Somatic and cranial dysplasia*: somatic or cranial asymmetry.
	6- Hydrocephalus.	6- Hydrocephalus.	
	7- Abnormalities of the connective tissue*.	7- Abnormal connective tissue*: hypermobility or hyperelastic skin	
	8-Asymmetry/macrosomia.		

***: sign present in our observation**