

Online Resource 5. Extra-osseous/extra-cutaneous abnormalities in CSHS published reports

Affected system	Number of subjects	Manifestations
Neurological abnormalities	21(45%)	<ul style="list-style-type: none"> • Intellectual disability/delayed milestones (10 subjects): [1-9] • Bilateral/unilateral ventricular enlargement (7 subjects): [3, 6, 8, 10-13] • Epilepsy/EEG abnormalities (5 subjects): [1, 2, 6, 8, 14] • Brainstem lipomas: (2 subjects): [13, 15] • Macrocephaly: (3 subjects): [13, 16, 17] • Brain atrophy (2 subjects):[8, 11] • Spastic hemiplegia (2 subjects): [8, 14] • Microcephaly (1 subject): [2] • Arachnoid cyst (2 subjects): [11, 18] • Partial third cranial nerve palsy (1 subject): [19]. • Hypoplastic right occipital bone (1 subject): [11]. • Dyslexia (1 subject): [13] • Nistagmus (1 subject): [15] • Hyperreflexia (1 subject):[15] • Interdigitation of gyri (1 subject): [20] • Brainstem tumor (non-lipoma) (1 subject):[21]
Ophthalmological abnormalities	12(%)	<ul style="list-style-type: none"> • Coloboma (5 subjects): [2, 13, 16, 17, 22] • Limbal dermoid (4 subjects): [8, 13, 17, 23] • Strabismus: (3 subjects): [2, 12, 24] • Corneal opacity: (3 subjects): [13, 22, 24] • Dermoid cyst of the eyelid: (1 subject): [16] • Scleral pigmented spots (1 subjects): [25]. • Ophthalmoplegia: (1 subject): [24] • Microcornea: (1 subject): [22] • Antimongoloid slant: (1 subject): [6] • Ptosis (1 subject): [6] • Corneal calcification (1 subject): [10]
Affected system	Number of subjects	Manifestations
Hemibody asymmetry	12 (%)	<ul style="list-style-type: none"> • Hemihypertrophy nevi side (4 subjects): [16, 26-28] • Hemihypoplasia nevi side: (5 subjects): [12, 14, 24, 29, 30] • Asymmetrical facies (2 subjects): [6]: side non-specified plus asymmetric dentition, [31]: tongue asymmetry (side non-specified)

Cardiovascular abnormalities	4 etc	<ul style="list-style-type: none"> Pericardial effusion (1 subjects): [23] (due to pulmonary hemangiomas). Aortic coarctation (1 subjects): [17] Heart valve abnormalities (1 subject): [10](mild aortic regurgitation due to calcification of aortic valve), Flat angioma (face)(1 subject): [26] Lymphedema (1 subject):[17]
Precocious puberty	4 etc	<ul style="list-style-type: none"> Central precocious puberty (3 subjects): [13, 19, 28] Not specified central/perif (1 subject): [24]
Thyroid abnormalities	1	<ul style="list-style-type: none"> Thyroid medullary carcinoma (1 subject): [32]: diagnosed at 15years, no family history)
Hyper IgE	2	<ul style="list-style-type: none"> [5, 33]
Hematological abnormalities	1	<ul style="list-style-type: none"> Severe anemia (unknown etiology) (1 subject): [34]
Basal cell carcinoma	2	<ul style="list-style-type: none"> Diagnose over 50 years: [24, 35]
Hyperparathyroidism	1	[33]
Congenital rhabdomyosarcoma	1	[28]
Testicular abnormalities	1	[17]
Giant cell granuloma of the maxilla	1	[16]
Pheocromo-cytoma	1	Detected incidentally at 54 [35]
Thymoma	1	<i>HRAS</i> mutation confirmed in timoma [31]
Affected system	Number of subjects	Manifestations
Bladder leiomyoma	1	[9]
NO extra-osseous/extra-cutaneous findings reported	15*	<ul style="list-style-type: none"> [1, 9, 10, 18, 36-43] *Not reported but obvious hemihypoplasia in images: [30]

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