

Multicenter study of long-term outcomes and quality of life in PHACE syndrome

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BACKGROUND

- PHACE syndrome describes the association between posterior fossa abnormalities, infantile hemangiomas (IH), arterial anomalies, cardiac anomalies, and eye anomalies. Long term outcomes have not been well described
- We aimed to investigate long-term outcomes and quality of life in individuals with definite PHACE ages ≥ 10 years

METHODS

- Multicenter, retrospective chart review and cross sectional participant interviews
- 16 sites contributed
- Investigators utilized standardized intake forms created by an expert panel

RESULTS

Table 1. Demographics and major clinical features (n=104)

Avg age	16.5 years
Median age	14 years
Age range	10-77 years
Female	89/104 (85.6%)
Indigenous	3/104 (2.9%)
Asian	7/104 (6.7%)
Black	4/104 (3.8%)
White	76/104 (73.1%)
Hispanic	21/104 (20.2%)
Facial IH	103/104 (99.0%)
Posterior fossa anomaly	51/104 (49.0%)
Arterial anomaly	96/104 (92.3%)
Cardiac anomaly	49/104 (47.1%)
Eye anomaly	20/104 (19.2%)

PHACE has multiple long-term morbidities including hemangioma residua (94.2%), headaches (72.1%), learning differences (45.1%), and progressive arteriopathy (29.4% of those with follow up imaging [n=68]) that call for specialized primary care and subspecialty follow up.

Figure 1. Patient reported headache frequency (left) and impact on daily life (right) (n=75)

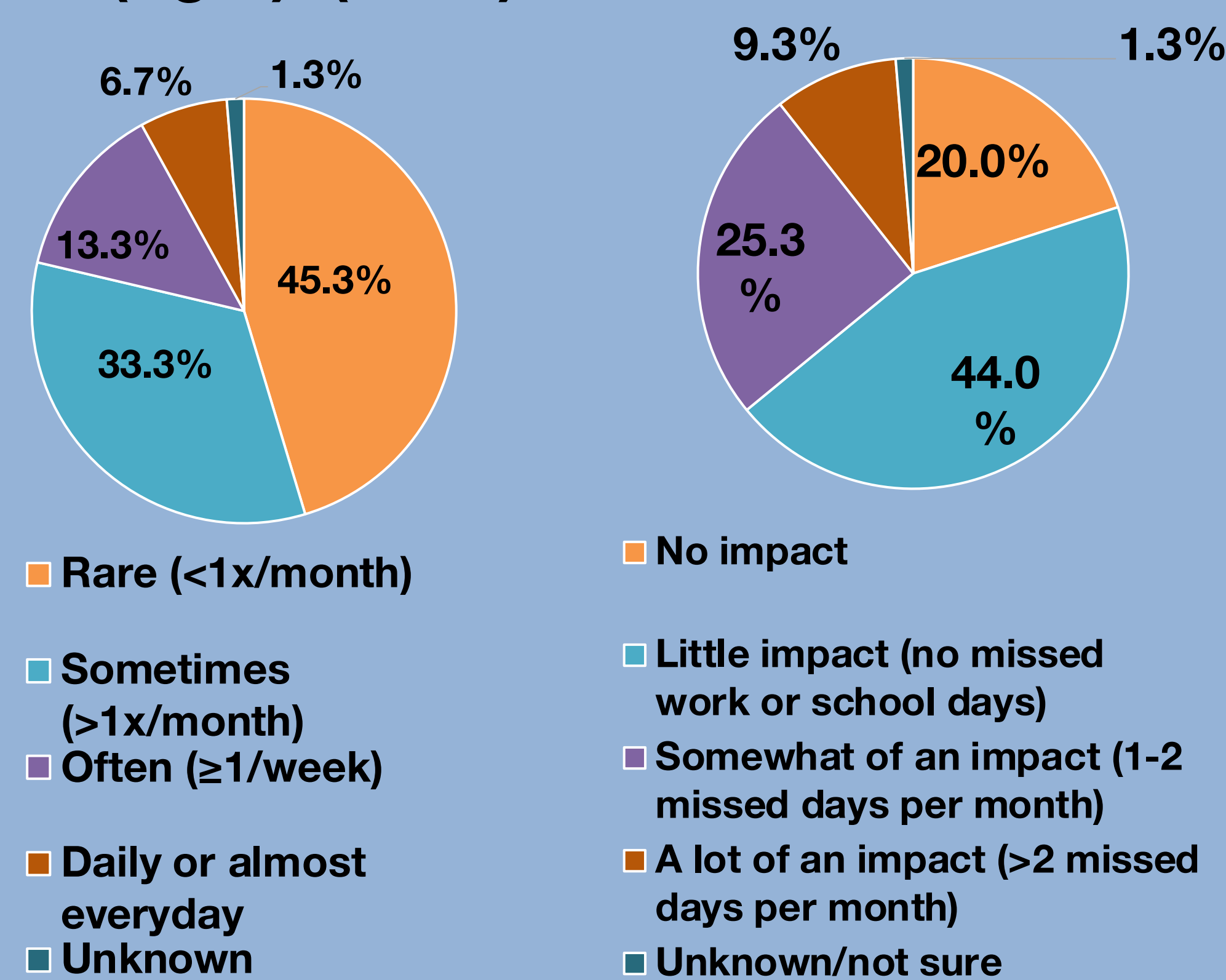


Figure 2. Patient reported satisfaction with IH appearance (n=104)

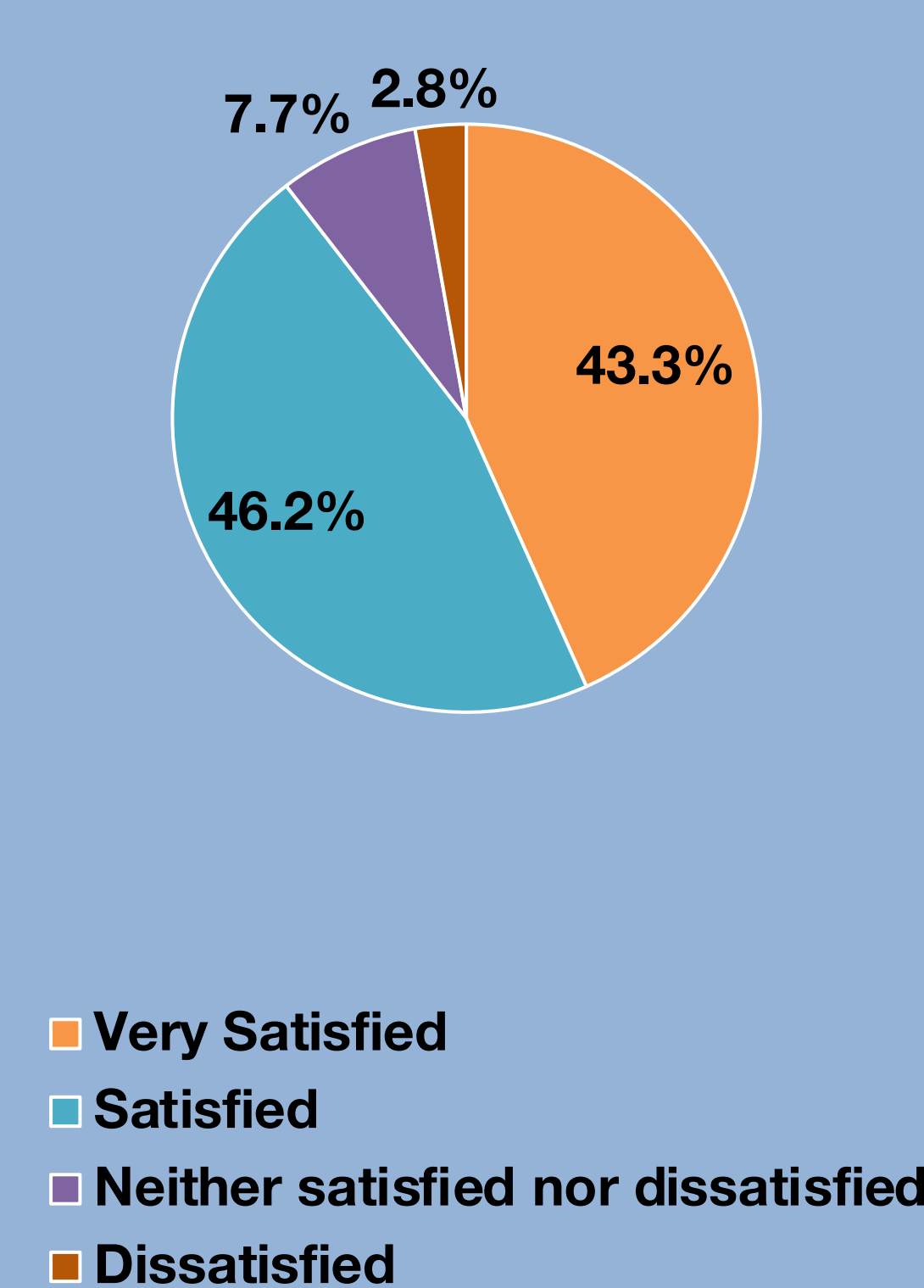


Table 3. Progressive arteriopathy (n=20)

Avg age	10.4 years
Median age	11.5 years
Age range	0.8-19 years
Low arterial risk	6/20 (30%)
Intermediate arterial risk	9/20 (15%)
High arterial risk	3/20 (15%)
Moyamoya (MM) syndrome	7/20 (35%)
Progressive stenosis/tortuosity without MM	11/20 (55%)
Progressive aneurysmal dilatation/ectasia	4/20 (20%)

Table 4. Logistic regression analyses

Impact on self-confidence	Surgical treatments: OR 0.145 (0.045-0.465) p=0.001 Ulceration in infancy: OR 0.291 (0.104-0.812)
Progressive arteriopathy	S4 IH location: OR 4.7 (1.303-16.6) p=0.018 Posterior fossa anomalies: OR 10.2 (2.5-41.4) p=0.0012
Learning differences	Posterior fossa anomalies: OR 4.0 (1.755-9.1) p=0.0010 Eye anomalies: OR 4.4 (1.433-13.5) p=0.0096
Headaches	IH size 10-15 cm: OR 5.5 (1.235-24.1) p=0.025 IH size > 50cm: OR 10.9 (1.194-99.7) p=0.034



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DISCUSSION

- Residual IH is common, but the majority were either very satisfied or satisfied with the appearance of the IH
- History of ulceration and surgical treatments in infancy were associated with an impact on patient reported self-confidence
- Headaches were seen in 72.1% (n=75). Of those who had headaches, 20% had ≥ 1 per week and 34.6% missed ≥ 1 day of school/work per month due to headaches
- Progressive arteriopathy occurred in 29% of the cohort and S4 IH location and posterior fossa anomalies were associated with increased risk
- Pediatric, parent proxy, and adult global health scores were moderately lower than population norms
- Limitations include ascertainment bias, evaluation of imaging reports only for details on progressive arteriopathies, and younger ages on average in the cohort

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