Genetic determinism and hemispheric influence in hair whorl formation

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Abstract

Background The mechanisms determining the laterality and the rotation direction of hair whorls are unknown. Here we report observations on twins investigating the genetic bases of whorl pattern formation. Knowing that vortex phenomena may depend on geographic effects, we also provide comparative data on whorls from children born in the Northern hemisphere (France) versus children born in the Southern hemisphere (Chile).

Material and methods We retrospectively included children from three populations: (1) Northern hemisphere general population, (2) Southern hemisphere general population, and (3) same-sex Northern hemisphere twins. We recorded whorl rotation direction (clockwise, counterclockwise), whorl position (left, right, central) and twinning type. Univariate logistic models were used to screen for associations between rotation direction and whorl position. For twins, the variable of interest was binary, i.e. same rotation direction (reference class) or opposite directions for each twin pair. For controls, all single combinations were included as virtual twins, and compared to real twins. Odds ratios (OR) were compared for both hemispheres, for real twins and virtual (control) twins.

Results Seventy-four (37 pairs) twins and 50 children from the general population of each hemisphere were included. The OR for opposite rotation directions between two twins was $\neq 1$ ($p=0.017$), meaning that whorls rotated preferentially in the same direction in twins. ORs were <1 for Northern and Southern hemispheres, meaning that whorls rotated preferentially in the same direction in simulated twins. OR was 0.04 [0.03; 0.05] for simulated twins in the Northern hemisphere, less than OR for real twins (0.42 [0.20; 0.83], $p < 0.001$), with no confidence interval superimposition, suggesting that it was more likely to observe opposite whorls in twins than in a random pair of children ($p < 0.001$). Also, OR for the Northern hemisphere (0.04 [0.03; 0.05]) was less than the OR for the Southern hemisphere (0.28 [0.24; 0.32]) with no confidence interval superimposition, which meant than counterclockwise whorls were more frequent in the Southern hemisphere ($p < 0.001$).

Conclusions We suggest that hair whorl formation is a genetically determined developmental process that can be influenced by extrinsic environmental factors. Our results furthermore underline the general importance of studies focused on limit phenomena that can provide insights on general developmental mechanisms. We plead for large-scale epidemiological assessments of hair whorls in several Northern and Southern hemisphere populations to confirm these surprising findings suggestion significant modulations of craniofacial development by geographic effects.

Introduction

According to the hedgehog theorem of algebraic topology [1], most hairy animals have hair whorls [2]: on full spheres, one cannot comb a hairy ball without creating a cowlick. In humans, hair whorls are constant on the scalp, even though this surface is a spherical cap rather than a full sphere. Their morphological characteristics are determined prenatally and there is currently no argument in favour of potential modifications during post-natal growth. Whorls can be unique or multiple, are generally lateralised, and oriented either clockwise or counterclockwise. Their developmental origins and the mechanisms determining their rotation direction are unclear. Both genetic and physical factors may be implicated in determining the location and orientation of whorls, as in other lateralisation phenomena [3]. As is Blaschko lines, which include whorl-like distributions on the scalp surface [4], hair whorls may potentially originate from cell migration processes but there is no current experimental evidence to support this hypothesis.

A report based on 404 new-borns showed that 98.5% of babies had single parietal whorls and 1.5% had double whorls, with 93.8% of clockwise whorls [2]. The most common configurations were, listed by decreasing prevalence: clockwise central, clockwise right, clockwise left, counterclockwise central, double, counterclockwise left, and counterclockwise right. Whorls have been associated with a surprising list of physical and cognitive characteristics: behaviour in cattle [5], laterality in humans [6] and even homosexuality [7] but all these reports have been invalidated [8, 9]. Several clinical arguments indicate that whorl formation may be - at least partially - under genetic control. For instance, atypical whorls (double or multiple, frontal hair whorls) seem to be more prevalent in patients with genetic conditions [10]. Twinning is a potential model to investigate the genetic bases of whorl distribution, by comparing monozygotic and dizygotic cases, even though monozygotic twins arise both from monochorionic pregnancies and from bichorionic pregnancies, with distinct mechanisms that could potentially interfere with whorl formation. Interestingly, whorls are regularly mentioned as a physical feature in mirror twins – representing 25% of monozygotic pairs – without supporting data in the literature. Furthermore, double occipital hair whorls seem to be more prevalent in twins than in singletons, and may be specifically prevalent in monozygotic twins [11].

In order to investigate the developmental origins of whorls, we performed an observational study in twins from monochorionic pregnancies (which are 100% monozygotic), and twins from bichorionic pregnancies of the same sex (which are 20% monozygotic and 80 % dizygotic) [12]. In order to evaluate environmental effects on whorl rotation direction, we compared whorl configuration in a population born in the Northern hemisphere (Paris, France) with a population born in the Southern hemisphere (Santiago, Chile). We obtained unexpected results.

Material and Methods

We conducted a retrospective study on three populations.

(1) Northern hemisphere general population, born in Paris, France, admitted for minor craniofacial trauma at Necker – Enfants Malades University Hospital (Paris, France), without diagnosed congenital conditions, from March 1st to March 31st 2021.

(2) Southern hemisphere general population, born in Santiago, Chile, admitted for minor craniofacial trauma at Clinica Universitad de los Andes (Santiago, Chile), without diagnosed congenital conditions, from March 1st to March 31st 2021.

(3) Same-sex Northern hemisphere twins - all pairs of same-sex twins born in Necker – Enfants Malades University Hospital (Paris, France) from January 1st 2022 to March 31st 2022.

children admitted for craniofacial trauma had minor facial lacerations and/or burns that had mostly occurred during household accidents, at school or during outdoors activities. The study was conducted according to the ethical rules for non-interventional investigations [13]. All patients benefited from clinical photography as part of their usual management and were informed of the conduct of the study.

Whorl rotation direction (clockwise, counterclockwise) and whorl position (left, right, central) were recorded by two independent examiners (MW, STL) and secondarily assessed by another author (RHK) without reported inconsistency. Twinning type: BiChorial BiAmniotic (BCBA), MonoChorial BiAmniotic (MCBA), and MonoChorial MonoAmniotic (MCMA) was extracted from medical files. As there is no current argument to suppose that whorl pattern may change with age, we did not include age into the list of variables and we considered that whorl characteristics at a given age corresponded to whorl characteristics at birth.

Univariate logistic models were designed for each explanatory variable to screen for a statistical association with rotation direction and position. For twins, the variable of interest was binary, i.e. same rotation direction (reference class) or opposite directions for each twin pair. For controls, all single children combinations were included as virtual twins, and compared to real twins. New odds ratios (OR) were calculated and compared for both hemispheres, for real twins and virtual (control) twins. The significance threshold was $p < 0.05$; a significant parameter influenced the relevant variables for each model. Assumptions of normality and homoscedasticity of errors were tested. The statistical analyses were performed on R 3.6.21 using the nlme2 and ggplot3 packages.

Results

Seventy-four twins were included. Absolute ratio values seemed to indicate that most whorls were clockwise and rotated in the same direction within pairs (Table 1). The OR for opposite rotation directions between two twins was $\neq 1$ ($p=0.017$), meaning that whorls rotated preferentially in the same direction in twins. The OR for different whorl laterality between two twins was not $\neq 1$ ($p=0.869$), meaning that whorls lateralised either on the same side or on opposite sides in twins. Sex, pregnancy type, number of chorions and whorl position did not affect rotation direction ($p<0.05$) and position ($p<0.05$).

In the general population, 50 children [25 girls and 25 boys], aged 1-10 were included in both Paris and Santiago centres. ORs were <1 for Northern and Southern hemispheres, meaning that whorls rotated preferentially in the same direction in simulated twins. Of note, OR was 0.04 [0.03; 0.05] for simulated twins in the Northern hemisphere, less than OR for real twins (0.42 [0.20; 0.83], $p < 0.001$), with no confidence interval superimposition, suggesting that it was more likely to observe opposite whorls in twins than in a random pair of children ($p < 0.001$). Also, OR for the Northern hemisphere (0.04 [0.03; 0.05]) was less than the OR for the Southern hemisphere (0.28 [0.24; 0.32]) with no confidence interval superimposition, which meant than counterclockwise whorls were more frequent in the Southern hemisphere ($p < 0.001$). All results are summarized in Table 2.

Discussion

Preferential rotation direction of hair whorls in twins from monochorionic pregnancies and same sex bichorionic pregnancies indicated that whorl characteristics were partially determined by genetic factors. A higher prevalence of counterclockwise whorls in the Southern hemisphere most probably indicated a potential role of environmental factors, even though genetic effects resulting from specific population characteristics could not be formally ruled out.

It is not clear whether this surprizing hemispheric effect on rotation direction could be related in any way to the Coriolis force. Classical experiments on liquid drainage show counterclockwise rotation in the Northern hemisphere [14] and clockwise rotation in the Southern hemisphere [15]. As multiple physical forces are involved in the initial steps of craniofacial development [16], there is probably no straightforward link between the rotation direction of whorls and any type of physical phenomenon potentially influenced by the Coriolis force, but this question requires further investigations.

We did not consider age as a factor that could potentially influence whorl morphological characteristics – this hypothesis cannot be formally ruled out even though it remains highly unlikely. Other non-hemispheric factors should thus be assessed on samples from various Northern and Southern hemisphere locations, such as maternal health, maternal nutrition, and/or prenatal hormone exposure, before considering a potential effect of hemispheric environmental physical factors such as the Coriolis force.

Conclusion

This report should lead to furthers investigations on the origins of hair whorls and more generally on the physical and chemical mechanisms determining the three-dimensional distribution of cell populations during development. Similarly, other enigmatic patterns such as concentric rings (Baló's concentric sclerosis, concentric skin rings in mycoses) or annular 'wood-grain' lesions (erythema gyratum repens) could provide critical information on general developmental processes by providing unique insights on phase transitions, critical points and the factors influencing order parameters [17].

Large-scale epidemiological assessments in genetically confirmed monozygotic twins and on both hemispheres in multiple locations are required to support our initial results and better understand the interactions between environmental factors and early developmental mechanisms.

If confirmed, these results indicate that hair whorl may be an indicator of the interactions between craniofacial development and extrinsic physical factors.

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Data availability statement : Data available within the article or its supplementary materials.

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Table 1. Data description. Total number of individuals: 74; total number of pairs: 37. BCBA = Bichorial Biamniotic; MCBA = Monochorial Biamniotic; Monochorial Monoamniotic.

		N (%)
Sex		
	Two girls	23/37 (62%)
	Two boys	14/37 (38%)
Type		
	BCBA	20/37 (54%)
	MCBA	16/37 (43%)
	MCMA	1/37 (3%)
Rotation direction		
	Total clockwise	64/74 (86%)
	Total anticlockwise	10/74 (14%)
	Same	26/37 (70%)
	Reverse	11/37 (30%)
Laterality		

	Total centered	38/74 (51%)
	Total left	14/74 (19%)
	Total right	22/74 (30%)
	Same	19/37 (51%)
	Reverse	18/37 (49%)

Table 2. A. Rotation direction in twins. Intercept value corresponded to the OR of opposite rotation directions between two twins. B. Laterality in twins. Intercept value corresponded to the OR of different whorl laterality between two twins. C. Control pairs. BCBA = Bichorial Biamniotic; MCBA = Monochorial Biamniotic; Monochorial Monoamniotic, BC = Bichorial; MC = monochorial.

A Opposite rotation direction		OR	IC 95%	p
Intercept		0.42	[0.20; 0.83]	0.017
Sex	Ref = Girls			
	Boys	2.70	[0.64; 12.1]	0.179
Type	Ref = BCBA			
	MCBA	1.62	[0.38; 7.50]	0.519
Chorion	Ref = MC			
	BC	0.57	[0.12; 2.37]	0.449
Whorl position	Ref = Same			
	Different	2.39	[0.57; 11.1]	0.241

B Different whorl laterality		OR	IC 95%	p
Intercept		0.95	[0.49; 1.81]	0.869
Sex	Ref = Girls			
	Boys	1.09	[0.29; 4.19]	0.898
Type	Ref = BCBA			
	MCBA	1.22	[0.33; 4.65]	0.765
Chorion	Ref = MC			
	BC	0.73	[0.19; 2.66]	0.630

Whorl position	Ref = Same			
	Different	2.39	[0.57; 11.1]	0.241

C Control pairs		OR	IC 95%	p
Intercept				
	Northern hemisphere	0.04	[0.03; 0.05]	< 0.001
	Southern hemisphere	0.28	[0.24; 0.32]	< 0.001
	Both populations	0.15	[0.14; 0.17]	< 0.001