

Genetic variation in personality traits explains genetic overlap between borderline personality features and substance use disorders

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ABSTRACT

Aims To examine the genetic overlap between borderline personality features (BPF) and substance use disorders (SUDs) and the extent to which variation in personality traits contributes to this covariance. **Design** Genetic structural equation modelling was used to partition the variance in and covariance between personality traits, BPF and SUDs into additive genetic, shared and individual-specific environmental factors. **Setting** All participants were registered with the Australian Twin Registry. **Participants** A total of 3127 Australian adult twins participated in the study. **Measurements** Diagnoses of DSM-IV alcohol and cannabis abuse/dependence (AAD; CAD) and nicotine dependence (ND) were derived via computer-assisted telephone interview. BPF and five-factor model personality traits were derived via self-report questionnaires. **Findings** Personality traits, BPF and substance use disorders were partially influenced by genetic factors with heritability estimates ranging from 0.38 (neuroticism; 95% confidence interval: 0.30–0.45) to 0.78 (CAD; 95% confidence interval: 0.67–0.86). Genetic and individual-specific environmental correlations between BPF and SUDs ranged from 0.33 to 0.56 (95% CI = 0.19–0.74) and 0.19–0.32 (95% CI = 0.06–0.43), respectively. Overall, there was substantial support for genetic influences that were specific to AAD, ND and CAD (30.76–68.60%). Finally, genetic variation in personality traits was responsible for 11.46% (extraversion for CAD) to 59.30% (neuroticism for AAD) of the correlation between BPF and SUDs. **Conclusions** Both genetic and individual-specific environmental factors contribute to comorbidity between borderline personality features and substance use disorders. A substantial proportion of this comorbidity can be attributed to variation in normal personality traits, particularly neuroticism.

Keywords Alcohol use disorders, borderline personality disorder, cannabis use disorders, comorbidity, five-factor model, genetics, nicotine dependence, personality traits, substance use disorders.

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Submitted 21 March 2014; initial review completed 15 May 2014; final version accepted 11 July 2014

INTRODUCTION

Borderline personality disorder is characterized by unstable relationships, identity disturbance, affective instability and impulsivity. Prevalence rates are estimated at 1.6–5.9% in the general population [1], and are considerably higher in out-patient and in-patient clinical settings (i.e. 10 and 20%, respectively). Additionally, it is associated with increased risk for comorbid physical [2] and mental health disorders [3]. One particularly prevalent

comorbidity is that between borderline personality disorder and substance use disorders (SUDs). Furthermore, individuals with borderline personality disorder are more likely to transition from asymptomatic to symptomatic substance use [4], persist in problematic use over a 3-year period [5] and are four to 10 times more likely to meet criteria for SUDs relative to those without a borderline diagnosis [6].

It is estimated that 40% of the variance in borderline personality disorder is accounted for by genetic factors

[7], while approximately 50% of the variance in SUDs is heritable [8]. Given this genetic underpinning, Bornovalova and colleagues [9] longitudinally examined genetic and environmental contributions to the comorbidity of borderline personality traits and substance use (i.e. mean quantity/frequency across alcohol, nicotine and marijuana) in adolescent twins, and demonstrated that shared environment accounted for all the associations between borderline personality and substance use at age 14 ($r_c = 1.00$); however, a moderate genetic correlation accounted for most of this association at age 18 ($r_g = 0.38$), suggesting that common genes influence these phenotypes. In adult twins, Distel and colleagues [10] examined genetic overlap between substance use and borderline personality features (BPF), which comprises affective instability, identity problems, negative relationships and self-harm derived from the Personality Assessment Inventory—borderline subscale [11]. They found that the correlation between BPF and both regular smoking and ever use of cannabis was explained by genetic factors ($r_g = 0.40$ and 0.51 , respectively), whereas unique environmental factors ($r_e = 0.32$) accounted for the overlap between BPF and high alcohol consumption. While these studies allude to shared genetic underpinnings between BPF and substance involvement, particularly during adulthood, neither study examined SUDs, a severe and more heritable manifestation of maladaptive substance involvement [12–15].

An additional gap in this literature is the role of personality traits in explaining the relationship between borderline personality disorder and SUDs. As incorporated into Section III of the 5th edition of the *Diagnostic and Statistical Manual* [1], personality disorders can be conceptualized as maladaptive configurations of personality traits and assessed using dimensional trait models, such as the five-factor model (FFM) [16]. Meta-analytical findings have demonstrated that FFM neuroticism (high), agreeableness (low) and conscientiousness (low) are associated most strongly with borderline personality disorder [17,18]. Furthermore, personality traits are heritable [19], and studies [20,21] have reported strong genetic correlations between BPF and neuroticism, conscientiousness, agreeableness and, in one case, extraversion [20]. More importantly, Distel and colleagues found that, when modelled simultaneously, genetic variation in these traits explains all the heritable variation in BPF. Therefore, personality traits may harbour important aetiological information regarding vulnerability to borderline personality disorder.

Meta-analytical research also shows that neuroticism, agreeableness and conscientiousness are linked to SUDs [22]. Specifically, significant effect sizes emerged between neuroticism and conscientiousness and alcohol and drug use disorders, as well as mixed SUDs (i.e. both alcohol and

drug). Low agreeableness was associated significantly with drug use and mixed SUDs only. Genetic variation in SUDs is also correlated with personality traits, in particular conscientiousness-related dimensions, as well as agreeableness- and extraversion-related dimensions [23–25].

The above findings suggest that FFM personality traits may be common aetiological risk factors for BPF–SUD symptomatology, and that variation in these traits may be partially implicated in the covariation between BPF and SUDs. Using data from a population-based sample of young adult twins, the current study examines (i) the role of overlapping genetic and environmental influences in the comorbidity between BPF and SUDs, including DSM-IV alcohol and cannabis abuse/dependence and nicotine dependence and, further, (ii) the extent to which genetic and environmental influences on FFM personality traits contribute to the covariation between BPF and SUDs.

MATERIALS AND METHODS

Sample

The initial sample consisted of 3348 twins, aged 21–40 years, from the Australian Twin Registry interviewed between 2005 and 2009 in a study focusing primarily on cannabis use [26]. Participants were interviewed using a computer-assisted telephone interview, and 3186 of the original sample completed a self-report questionnaire, on average, within 2 weeks of the interview. The final sample included 3127 twins with complete data for relevant study variables (i.e. SUDs, BPF and FFM traits): (i) 948 monozygotic female twins (MZF; 382 pairs, 184 unpaired); (ii) 444 monozygotic male twins (MZM; 153 pairs, 138 unpaired); (iii) 716 dizygotic female twins (DZF; 288 pairs, 140 unpaired); (iv) 330 dizygotic male twin (DZM; 95 pairs, 140 unpaired); and (v) 689 dizygotic opposite sex twins (DZOS; 198 pairs, 293 unpaired). Mean age in the current study was 31.84 [standard deviation (SD) = 2.48].

Measures

SUDs

SUDs were assessed using the Australian version of the Semi-Structured Assessment of the Genetics of Alcoholism (SSAGA) (Australian version; SSAGA–OZ), which utilizes a computer-administered telephone interview. The SSAGA [27] has demonstrated good reliability and validity as a measure of standardized DSM-IV diagnostic criteria for a range of conditions including SUDs [28]. For the current analyses, binary variables were created for DSM-IV diagnoses of alcohol and cannabis abuse/

dependence (AAD and CAD, respectively) and nicotine dependence (ND). For AAD and CAD, a '1' was coded if participants met criteria for either abuse or dependence and a '0' for neither abuse nor dependence. For AAD, CAD and ND, prevalence rates in the current sample were 47.2, 15.6 and 25%, respectively.

BPF

BPF were assessed by the 24-item, self-reported Personality Assessment Inventory–borderline subscale (PAI–BOR [11]). The PAI–BOR generates a total score and scores on four subscales. In the current study, only the total score was used ($\alpha = 0.88$). PAI–BOR scores were divided into five approximately equal categories and tested for multivariate normality prior to phenotypical and twin modelling analyses.

FFM

FFM personality traits were assessed using the 60-item, self-report NEO five-factor inventory (NEO–FFI; [29]). Items are scored on a 1 (strongly disagree) to 5 (strongly agree) scale, and domain scores were generated by computing the mean of items from each subscale. Alphas for neuroticism, extraversion, openness, agreeableness and conscientiousness were 0.89, 0.80, 0.73, 0.76 and 0.84, respectively. Each domain score was divided into quartiles and tested for multivariate normality prior to phenotypical and twin modelling analyses.

Data analysis

Phenotypical analyses (i.e. correlations between BPF, FFM personality traits and SUDs) were conducted in SAS. Only FFM traits correlated statistically significantly with both BPF and SUDs (at $P < 0.05$) were included in twin modelling procedures, conducted in Mx [30] using raw ordinal data and full information maximum likelihood (FIML) estimation. A series of trivariate Cholesky models were fitted to the data (Fig. 1). Each FFM personality trait was modelled first, followed by BPF, and then each SUD. Initial analyses allowed for differing thresholds (adjusted for age) and separate estimates for additive genetic (a^2), shared environmental (c^2) and individual-specific environmental (e^2) influences on male and female twins. An omnibus test of sex differences was utilized to investigate whether constraining variance components (a^2 , c^2 and e^2), but not thresholds, across sexes resulted in a significant deterioration of fit using the difference in -2 log-likelihood fit, which is distributed as a χ^2 . Similarly, the role of c^2 was examined by constraining it to zero. Due to the relatively small sample size and uneven distribution of same- versus opposite-sex DZ pairs, qualitative sex differences ($r_{DZOS} < > 0.5$) were not explored. In addition to attributing variance in FFM personality, BPF and SUDs to

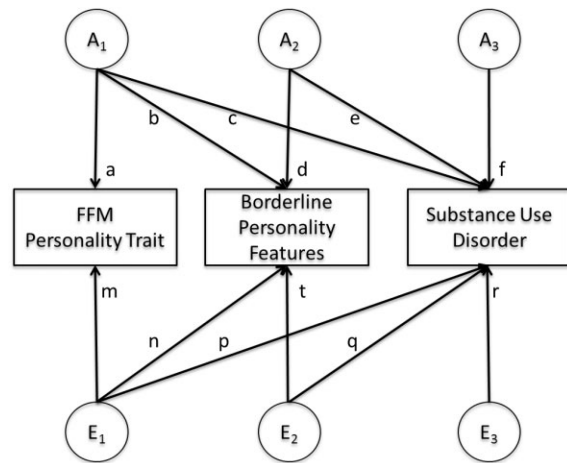


Figure 1 Trivariate Cholesky decomposition model. A_1 = genetic influences shared in common by five-factor model (FFM) trait, borderline personality features (BPF) and substance use disorder (SUD). A_2 = genetic influences shared in common by BPF and SUD. A_3 = genetic influences specific to SUD. E_1 = individual-specific environmental influences shared in common by five-factor model (FFM) trait, borderline personality features (BPF) and substance use disorder (SUD). E_2 = individual-specific environmental influences shared in common by BPF and SUD. E_3 = individual-specific environmental influences specific to SUD

a^2 , c^2 and e^2 , we calculated the extent to which variance in SUDs and covariance between them and BPF was attributable to genetic and environmental influences on FFM personality.

RESULTS

Phenotypical correlations

Polychoric and tetrachoric correlations between BPF, FFM personality traits and SUDs are shown in Table 1 ($n = 3127$). Strong correlations were noted between BPF and FFM traits, particularly neuroticism (0.75) and agreeableness (-0.48). The correlation between openness and BPF was non-significant, and openness was excluded from subsequent genetic analyses. BPF was correlated moderately with all three SUD variables (AAD = 0.25, CAD = 0.30, ND = 0.36). The SUDs were intercorrelated moderately to strongly (0.40–0.60). All correlations between SUDs and FFM traits were significant, except for openness and ND (0.03) and extraversion and AAD (0.04). The latter relationship was therefore not explored from a genetic perspective. The remaining correlations ranged from $|0.07|$ (openness and AAD) to $|0.25|$ (conscientiousness and CAD). Based on these phenotypical associations, we examined 11 genetic models (i.e. the extent to which each of the four FFM personality traits accounted for the correlation between BPF and the three SUD measures, excluding extraversion and AAD). This trivariate model is illustrated in Fig. 1.

Table 1 Phenotypical correlations and 95% confidence limits between borderline personality features, five-factor model personality traits, and substance use variables.

	BPF	N	E	O	A	C	AAD	CAD	ND
BPF	–								
N	0.75* (0.73–0.77)	–							
E	–0.36* (–0.40 to –0.33)	–0.51* (–0.54 to –0.47)	–						
O	0.00 (–0.04 to 0.04)	–0.02 (–0.06 to 0.02)	0.15* (0.11 to 0.18)	–					
A	–0.48* (–0.52 to –0.46)	–0.38* (–0.41 to –0.35)	0.30* (0.26 to 0.33)	0.09* (0.05 to 0.13)	–				
C	–0.36* (–0.40 to –0.32)	–0.41* (–0.45 to –0.38)	0.33* (0.29 to 0.36)	–0.02 (–0.06 to 0.03)	0.30* (0.27 to 0.34)	–			
AAD	0.25* (0.20 to 0.29)	0.15* (0.11 to 0.20)	0.04 (–0.01 to 0.09)	0.07* (0.03 to 0.12)	–0.19* (–0.24 to –0.14)	–0.15* (–0.20 to –0.10)	–		
CAD	0.30* (0.25 to 0.036)	0.15* (0.09 to 0.21)	–0.10* (–0.16 to –0.04)	0.14* (0.08 to 0.20)	–0.21* (–0.26 to –0.15)	–0.25* (–0.30 to –0.19)	0.48* (0.42 to 0.53)	–	
ND	0.36* (0.32 to 0.41)	0.23* (0.18 to 0.28)	–0.12* (–0.17 to –0.07)	0.03 (–0.02 to 0.09)	–0.20* (–0.25 to –0.15)	–0.20* (–0.25 to 0.15)	0.40* (0.34 to 0.45)	0.60* (0.54 to 0.65)	–

* $P < 0.05$; BPF = borderline personality features; N = neuroticism; E = extraversion; O = openness; A = agreeableness; C = conscientiousness; AAD = life-time alcohol abuse or dependence; CAD = life-time cannabis abuse or dependence; ND = life-time nicotine dependence; $n = 3127$.

Twin modelling

All polychotomous variables satisfied the assumption of multivariate normality ($P_s > 0.05$). All results are shown for models where parameters were constrained to be equal across male and female twins, as there was no significant evidence for sex differences ($P > 0.05$). Univariate heritability estimates (a^2), ranging from 0.38 (neuroticism) to 0.78 (CAD), for BPF, FFM traits and the SUDs are shown in Table 2 ($n = 3127$). The remainder of the variance in each measure, except for ND, could be attributed to individual-specific environmental factors (e^2). While shared environmental influences could not be constrained to zero for ND, for all other measures such a constraint did not produce a significant deterioration in model fit (χ^2 ranging from 3.173 to 4.985, d.f. = 6, $P > 0.05$).

Genetic (r_g) and environmental (r_e) correlations between BPF and FFM traits as well as SUDs are shown in Table 3 ($n = 3127$). The strongest genetic and environmental overlap with BPF emerged for neuroticism, followed by agreeableness. The phenotypical correlation between BPF and AAD, ND and CAD was attributable to genetic and environmental influences as well, with genetic correlations ranging from 0.33 to 0.56 and individual-specific environmental correlations ranging from 0.19 to 0.32. Genetic correlations between FFM traits and SUDs (not shown in Table 3) ranged from $|0.11|$ (extraversion and CAD) to $|0.46|$ (agreeableness and ND); individual-specific environmental correlations ranged from $|0.03|$ (agreeableness and AAD) to $|0.29|$ (neuroticism and CAD).

Importantly, we were interested in the proportion of variance in SUDs attributable to genetic and environmental influences on BPF and FFM personality traits—we provide proportions of the total variance in Table 4 ($n = 3127$) and, individually, proportions of genetic and individual-specific environmental variance separately in Supporting Information Table S1. Overall, there was substantial support for genetic influences specific to AAD, ND and CAD that were unshared with BPF and FFM personality traits (30.76–68.60%). Genetic influences on FFM traits contributed from 2.21% (neuroticism and CAD; the overlap between extraversion and CAD was not significant) to 9.75% (agreeableness and ND) of the total variance in SUDs (approximating 2.97–21.14% of the genetic variance; Supporting Information Table S1). Even after accounting for that overlap and genetic overlap between FFM traits and BPF (Table 3), approximately 1.71% (conscientiousness and AAD; the overlap between neuroticism and AAD was not significant) to 12.48% (neuroticism and ND) of the total variance in SUDs was attributable to BPF (approximating 3.28–22.61% of the genetic variance; Supporting Information Table S1).

Table 2 Standardized estimates of additive genetic (a^2), shared (c^2) and individual-specific (e^2) environmental variance for borderline personality features, five-factor model personality traits and substance use disorders.

	a^2 (95% CI)	c^2 (95% CI)	e^2 (95% CI)
Borderline personality features (BPF)	0.49 (0.42–0.55)	–	0.51 (0.45–0.58)
Neuroticism (N)	0.38 (0.30–0.45)	–	0.62 (0.55–0.70)
Extraversion (E)	0.41 (0.33–0.48)	–	0.59 (0.52–0.67)
Agreeableness (A)	0.40 (0.32–0.48)	–	0.60 (0.52–0.68)
Conscientiousness (C)	0.42 (0.34–0.49)	–	0.58 (0.50–0.66)
Alcohol abuse/dependence (AAD)	0.54 (0.43–0.64)	–	0.46 (0.36–0.57)
Cannabis abuse/dependence (CAD)	0.78 (0.67–0.86)	–	0.22 (0.14–0.33)
Nicotine dependence (ND)	0.47 (0.17–0.77)	0.27 (0.00–0.51)	0.26 (0.18–0.36)

All estimates are statistically significant at $P < 0.05$; $n = 3127$. CI = confidence interval.

Table 3 Genetic (r_g) and individual-specific environmental correlations (r_e) between borderline personality features (BPF) and substance use disorders (SUDs), accounting for correlations between BPF and five-factor model (FFM) personality traits (shown) and FFM and SUDs (not shown).

	Borderline personality features	
	r_g (95% CI)	r_e (95% CI)
Neuroticism (N)	0.87 (0.80 to 0.93)	0.68 (0.62 to 0.73)
Extraversion (E)	–0.52 (–0.63 to –0.40)	–0.22 (–0.31 to –0.13)
Agreeableness (A)	–0.65 (–0.74 to –0.54)	–0.39 (–0.46 to –0.30)
Conscientiousness (C)	–0.45 (–0.51 to –0.34)	–0.26 (–0.34 to –0.17)
Alcohol abuse/dependence (AAD)	0.33 (0.19 to 0.47)	0.19 (0.06 to 0.32)
Cannabis abuse/dependence (CAD)	0.34 (0.20 to 0.46)	0.32 (0.14 to 0.37)
Nicotine dependence (ND)	0.56 (0.39 to 0.74)	0.28 (0.13 to 0.43)

All estimates are statistically significant at $P < 0.05$; $n = 3127$. CI = confidence interval.

Broadly, speaking, environmental influences on FFM personality traits did not overlap consistently with those impacting SUDs (with a few exceptions, i.e. neuroticism for CAD and ND), but there was consistent although modest overlap in individual-specific environmental influences on BPF and SUDs.

Table 5 shows similar estimates, but for the proportion of total covariance between BPF and SUDs due to FFM traits ($n = 3127$). Proportions of covariance between BPF and SUD accounted for by personality computed individually for genetic and individual-specific environmental covariance are presented in Supporting Information Table S2. FFM personality traits were responsible for 14.55% (extraversion for ND) to 59.30% (neuroticism for AAD) of the total correlation/covariance between BPF and SUDs. With the exception of neuroticism (10.67–21.34%), the total covariance between BPF and SUDs was largely independent of individual-specific environmental influences on personality, in that the covariance accounted for by extraversion, agreeableness, and conscientiousness ranged from 0.81% (conscientiousness and ND) to 4.89% (conscientiousness and CAD).

As we were most interested in the extent to which genetic influences contributed to the relationship between individual SUDs, BPF and FFM personality, our genetic results (Supporting Information Tables S1 and S2) are summarized by SUD below.

AAD

Genetic variance in AAD was derived partially from neuroticism, agreeableness and conscientiousness, as well as from BPF, but only in the absence of neuroticism. In fact, the genetic covariance between AAD and BPF was entirely attributable to genetic overlap with neuroticism. However, agreeableness and conscientiousness only explained a proportion of the genetic covariance between BPF and AAD.

ND

The genetic variance in ND overlapped most considerably with BPF and agreeableness, with the latter responsible for 53% of the covariance between BPF and ND. The personality traits of extraversion and conscientiousness contributed genetic influences to ND and the covariance

Table 4 Percentage of total variance in substance use disorders attributable to genetic and environmental variation in five-factor model personality traits, borderline personality features (BPF) and specific to substance use disorder (SUD).

<i>Additive genetic factors</i>				
	<i>Personality</i>		<i>BPF</i>	<i>Specific</i>
	%		%	%
Neuroticism				
AAD	6.58		0.10	NS 46.38
CAD	2.28		11.40	63.01
ND	6.35		10.99	30.76
Extraversion				
AAD	–		–	–
CAD	1.00	NS	7.88	68.54
ND	2.21		12.48	32.22
Agreeableness				
AAD	3.88		2.17	47.22
CAD	3.40		5.51	68.60
ND	9.75		5.41	30.96
Conscientiousness				
AAD	6.09		1.71	44.41
CAD	3.65		5.14	68.06
ND	7.06		8.56	32.13
<i>Individual-specific environmental factors</i>				
	<i>Personality</i>		<i>BPF</i>	<i>Specific</i>
	%		%	%
Neuroticism				
AAD	0.32	NS	1.60	45.04
CAD	1.95		0.69	NS 20.67
ND	1.65		0.62	NS 24.19
Extraversion				
AAD	–		–	–
CAD	0.38	NS	2.09	20.11
ND	0.11	NS	2.03	24.58
Agreeableness				
AAD	0.06	NS	1.74	44.93
CAD	0.05	NS	2.50	19.92
ND	0.14	NS	3.20	23.76
Conscientiousness				
AAD	0.17	NS	2.44	45.19
CAD	0.71	NS	2.12	20.31
ND	0.03	NS	2.32	24.17

NS = not significant at $P < 0.05$; all other estimates are significant at $P < 0.05$; AAD = life-time alcohol abuse or dependence; CAD = life-time cannabis abuse or dependence; ND = life-time nicotine dependence; from Fig. 1, total variance in SUDs = $c^2 + e^2 + f^2 + p^2 + q^2 + r^2$ (e.g. genetic % personality = $c^2/c^2 + e^2 + f^2 + p^2 + q^2 + r^2$; environmental % personality = $p^2/c^2 + e^2 + f^2 + p^2 + q^2 + r^2$); For ND, added genetic and individual-specific percentages do not equal 100% due to significant contribution of shared environmental factors, which are not shown in this table; $n = 3127$.

between BPF and ND to a more modest degree. However, while neuroticism contributed to only 13% of the genetic variance in ND, it was responsible for more than half the covariance between BPF and ND.

Table 5 Proportion of total covariance between borderline personality features (BPF) and substance use disorders (SUD) explained by genetic and environmental variation in five-factor model personality traits and specific to variation shared by BPF and SUD.

	<i>A</i>		<i>E</i>	
	% <i>personality</i>	% <i>specific</i>	% <i>personality</i>	% <i>specific</i>
Neuroticism				
AAD	59.30	4.29	10.67	25.75
CAD	28.54	36.39	21.34	13.74
ND	40.81	31.00	16.98	11.20
Extraversion				
AAD	–	–	–	–
CAD	11.46	53.54	3.09	31.92
ND	14.55	57.14	1.43	26.87
Agreeableness				
AAD	34.15	30.23	2.50	33.12
CAD	26.15	39.58	1.94	32.34
ND	35.72	31.88	2.63	29.78
Conscientiousness				
AAD	28.70	29.23	2.73	39.34
CAD	18.89	44.40	4.89	31.83
ND	22.65	48.22	0.81	28.32

AAD = life-time alcohol abuse or dependence; CAD = life-time cannabis abuse or dependence; ND = life-time nicotine dependence; A = genetic covariance; E = individual-specific environmental covariance. From Fig. 1, A% personality = $bc/(bc + de + np + tq)$; A% specific = $de/(bc + de + np + tq)$; E% personality = $np/(bc + de + np + tq)$; E% specific = $tq/(bc + de + np + tq)$; $n = 3127$.

CAD

While BPF contributed modestly to the variance in CAD, the proportion of variance in CAD attributable to FFM personality traits was very small (0.4–4.8%). The traits of neuroticism and agreeableness contributed more substantially (40–43%) to the covariance between BPF and CAD than extraversion and conscientiousness (29–30%).

DISCUSSION

The current study sought to parse the genetic and environmental contributions to comorbid BPF and SUDs, as well as the extent to which variation in FFM traits explains this overlap. As expected, BPF was correlated with all three SUD variables, and BPF and SUDs were associated most consistently with neuroticism, agreeableness and conscientiousness. The correlation between BPF and neuroticism ($r = 0.75$) is somewhat higher than that reported in meta-analytical research [18]. However, a strong correlation is expected, given that personality disorders are conceptualized as maladaptive configurations of normal traits, and neuroticism is most relevant to

the borderline diagnosis. This high correlation could reflect common method variance or semantic overlap, although Distel and colleagues [20] indicated that 'removing systematically one of the 12 items of the neuroticism scale does not result in a large change in the correlation with the PAI-BOR, thus the influence of semantic overlap on the association is unlikely to be large' (p. 1136). The correlation between BPF and extraversion was also significant (-0.36), which is inconsistent with meta-analytical research [17,18]; however, the latter did not include studies using the PAI to assess borderline pathology, and previous research using the PAI has shown that individuals scoring highly on BPF report significantly lower scores on FFM extraversion [31]. The non-significant correlation between extraversion and AAD might also be considered surprising, given that sensation-seeking is a core component of this personality domain and linked consistently to alcohol use [32,33]. This may reflect a limitation of the NEO-FFI, in that only one item assesses the excitement-seeking facet. However, meta-analysis has revealed non-significant effect sizes between extraversion and substance use pathology [22].

In general, the magnitude of additive genetic and individual-specific environmental contributions to BPE, SUDs and FFM traits were highly consistent with the published literature [7,8,19]. Although we used ordinal FFM variables, estimates of heritability were nearly identical to studies that have used continuously distributed scores [19]. The lack of the role of shared environment also mirrors published work. We were, however, unable to constrain shared environmental influences on ND to zero, which departs from previous findings [15,34].

The primary aim was to examine genetic and environmental contributions to the comorbidity between BPF and SUDs. Moderate genetic correlations emerged between BPF and SUDs (i.e. 0.33, 0.34 and 0.56 for AAD, CAD and ND, respectively). Other studies examining comorbid heritability of BPF and substance involvement also reported moderate genetic influences for life-time cannabis use and regular tobacco use in adults, as well as with tobacco, alcohol and cannabis use at age 18 [9,10]. However, Distel and colleagues reported that only the individual-specific environmental correlation was significant between BPF and high alcohol consumption (i.e. 21+ drinks/week for males; 14+ drinks/week for females), whereas the current results indicate that comorbid AAD and BPF are influenced by both individual-specific environmental and genetic factors. This may reflect differences in the importance of genetic factors at various stages of substance use, and extant research [12–15] supports the notion that later stages of involvement (e.g. SUDs) may be more heavily genetically influenced than earlier stages (e.g. initiation, regular use).

Genetic variation in personality traits has also been implicated in the aetiology of substance involvement, albeit to a lesser degree. This study provides the most comprehensive examination of the genetic overlap between FFM traits and SUDs. Small to moderate genetic correlations emerged for the overlap between FFM traits and both AAD and ND ($|0.22|$ – $|0.46|$). More modest genetic correlations emerged for CAD and FFM traits ($|0.17|$ to $|0.22|$). These findings are generally in line with two studies examining FFM personality traits and alcohol involvement—one found small to modest significant phenotypical correlations between all five FFM domains and problem drinking [35,35]. An additional study reported moderate genetic correlations between alcohol use disorder symptoms and neuroticism, agreeableness and conscientiousness, but reported that drinking motives accounted for this genetic overlap [24]. The findings for cannabis are nearly identical to genetic correlations reported previously between CAD and neuroticism and extraversion [23]. There is a dearth of research, however, regarding the correlation between tobacco involvement and FFM traits. For instance, Heath and colleagues [36] examined the correlation between neuroticism and extraversion drawn from the Eysenck Personality Questionnaire and tobacco smoking and found the associations too modest to decompose into genetic and environmental sources. Overall, findings in the current study are consistent with molecular genetic studies suggesting that personality traits and SUDs, in part, share a genetic basis [37,38] and, furthermore, that personality traits may partially mediate the relationship between specific genetic polymorphisms and SUDs [39].

Finally, we examined the extent to which variation in personality traits explains the comorbidity between BPF and SUDs. Overall, genetic variation in FFM traits accounted for a substantial portion of the genetic covariance between BPF and SUDs, and in one case, variation in neuroticism completely explained the genetic comorbidity between BPF and AAD. This is consistent with theoretical and empirical work conceptualizing personality disorders as maladaptive configurations of normal personality traits [40] and provides support for the notion that personality trait variation may explain patterns of psychiatric comorbidity [41]. Specifically, researchers have suggested that traits such as affective lability and impulsivity are a common diathesis for borderline personality disorder and SUDs and thus contribute to their co-occurrence [42]. However, there is somewhat limited coverage of these traits within the NEO-FFI, given that it assesses only the FFM domains (and not facet-level traits), which probably attenuated the proportion of genetic covariance in BPF–SUD accounted for by the FFM traits. Generally, these findings have

important implications, because identifying aetiological mechanisms of risk for psychopathology can inform more effective treatment. Specifically, these data suggest that personality traits may represent an intermediate phenotype in the risk for comorbid BPF and SUD. Therefore, greater attention in addiction genetics research to identification of genes conferring vulnerability for personality traits may be fruitful. Also, interventions targeting more basic traits that give rise to various forms of psychopathology (e.g. affective instability), rather than disorder-specific interventions, may be more effective in prevention and treatment.

Limitations beyond the lack of facet-level FFM trait assessment include the use of self-reported BPF rather than interview-derived data. Additionally, the study sample size limited the ability to model FFM traits simultaneously in relation to BPF and SUDs. For instance, the FFM personality trait domains share genetic underpinnings and a multivariate model that accounts, simultaneously, for this overlap as well as that between these traits, BPF and SUDs may be informative in a larger sample. Similarly, modelling the genetic overlap simultaneously across the three SUDs could be conducted, and may be warranted given the high intercorrelations among the SUD variables. A larger sample would also allow disentangling the extent to which the genetic overlap between BPF, FFM personality traits and SUDs is due to covariation with earlier stages of substance experimentation and regular use versus later stages of abuse/dependence.

In conclusion, results of the current study indicate that both genetic and individual-specific environmental factors contribute to comorbid BPF and SUDs. More importantly, genetic variation in normal personality traits accounts for a substantial proportion of the genetic covariance between BPF and SUDs, particularly for neuroticism and AAD. These findings support the notion that personality traits confer genetic vulnerability to comorbid BPF and SUDs. This is consistent with theoretical and empirical work that influenced the inclusion of a dimensional personality trait model in Section III of the DSM-5 [1]. Not only can this trait model be utilized in the assessment of personality disorders, it could also be used as a hierarchical framework for classifying specific disorders in the DSM [41]. Specifically, Krueger and colleagues suggest that personality traits (e.g. disinhibition) represent the core of various spectra of psychopathology that could be organized in the DSM based on commonly observed patterns of comorbidity (e.g. externalizing disorders). These are important considerations for future iterations of the DSM and, moving forward, research examining borderline personality disorder and comorbid psychopathology should account for the role of personality traits.

Declaration of interests

This research was funded by National Institute on Drug Abuse (NIDA) grants. DA18267 (ML; data collection); DA23668 and K02DA32573 (AA; analysis) and T32DA007313 (LF); and facilitated through access to the Australian Twin Registry, a national resource supported by an Enabling Grant (ID 628911) from the National Health and Medical Research Council. A.A. has received peer-reviewed grant funding, travel reimbursements and an honorarium from ABMRF/Foundation for Alcohol Research, which receives some of its funds from brewers.

Acknowledgements

We thank Anjali Henders, Richard Parker, Soad Hancock, Judith Moir, Sally Rodda, Pieta-Maree Shertock, Heather Park, Jill Wood, Pam Barton, Fran Husband and Adele Somerville, who worked on this project, and the twins and their siblings for participating.

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Supporting information

Additional Supporting Information may be found in the online version of this article at the publisher's web-site:

Table S1 Proportion of additive genetic and individual-specific environmental variance in substance use

disorders attributable to five-factor model personality traits, borderline personality features and specific to substance use disorder.

Table S2 Proportion of genetic covariance between borderline personality features and substance use disorder explained by five-factor model personality traits.