

User Manual

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1 Introduction

E-Pedigrees: a large-scale automatic family pedigree prediction application which was developed as a novel fully automated software to construct family pedigrees from information readily available in an EHR system [Huang et al., 2021]. *E-pedigrees* infers familial relationships using two previously published prediction algorithms including Family Pedigree Prediction Algorithm (**FPPA**) [Huang et al., 2017] and Relationship Inference from the Electronic Health Record (**RIFTEHR**) [Polubriaginof et al., 2018].

2 Usage:

Please follow the exact input format for all the input files.

2.1 FAAP:

run *E-pedigrees*: python main.py FPPA

Enter your input files for **FPPA**: address.csv name.csv demo.csv account.cs pc.txt familyTree.csv

Enter one PED file if any: ped.csv [optional] (you can leave it blank)

2.2 RIFTEHR

run *E-pedigrees*: python main.py RIFTEHR

Enter your input files for **RIFTEHR**: patient.csv ec.csv familyTree.csv

Enter one PED file if any: ped.csv [optional] (you can leave it blank)

2.3 Both:

run *E-pedigrees*: python main.py both

Enter your input files for **BOTH**: address.csv name.csv demo.csv account.cs pc.txt patientc.csv ec.csv familyTree.csv

Enter one PED file if any: ped.csv [optional] (you can leave it blank)

3 Input Files

3.1 FPPA

Input files for address file in table 1, name file in table 2, demographic file in table 3, account file in table 4

3.2 RIFTEHR

Input files for patient file in table 5, and emergency contact file in table 6.

study_id	street_1	street_2	city	state	zip	from_year	thru_year
1	790393		7200	28	18216		
10	117141		5115	28	11753		2005
56	221591	448275	2893	28	9427	2003	2011

Table 1: Address information file format.

study_id	last_name_id	first_name_id	middle_name_id	from_year	thru_year
1	103775	53806			
10	46972	44623		2005	2011
50	2696	62099		1997	2007
50	105616	62099			1997

Table 2: Name information file format.

study_id	gender_code	birth_year	deceased_year	PHONE_NUM_id	from_year	thru_year
1	F	1989				
2	F	1947		134271		2011
282056	U	1986	2010			

Table 3: Demographic information file format.

study_id	ACCT_NUM_id	from_year	thru_year
2	982162		2011
10	523063	2005	2011

Table 4: Account information file format.

PatientID	FirstName	LastName	Sex	PhoneNumber	Zipcode	birth_year	deceased_year
1	103775	53806	M	1112223333	18216	1970	
10	46972	44623	M	2223334444	11753	1972	
50	2696	62099	F	3334445555	18216	1980	
96	105616	53806	F	1112223333	10032	1956	
122	345228	44623	F	2223334444	11753	1990	

Table 5: Patient information file format.

PatientID	EC_FirstName	EC_LastName	EC_PhoneNumber	EC_Zipcode	EC_Relationship
1	105616	53806	1112223333	18216	Mother
10	345228	44623	2223334444	11753	Father

Table 6: Emergency contact file format.

family_ID	num_fam_member	individual_ID	Maternal_ID	Paternal_ID	Gender
1	5	50	1112223333	18216	M
2	3	96	2223334444	11753	F

Table 7: Pedigree file format.

3.3 PED file

Pedigree file format in table 7.

References

- Xiayuan Huang, Nicholas Tatonetti, Katie LaRow, Brooke Delgoﬀee, John Mayer, David Page, and Scott J Hebbring. E-Pedigrees: a large-scale automatic family pedigree prediction application. *Bioinformatics*, 37(21):3966–3968, 06 2021. ISSN 1367-4803. doi: 10.1093/bioinformatics/btab419. URL <https://doi.org/10.1093/bioinformatics/btab419>.
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