

Annotation Enhancement of A Synthetic Family History Corpus

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Aunt

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Background

- As a key element for precision medicine, family history (FH) remains a challenge to obtain from unstructured clinical texts. To overcome the limitation by difficulties to access annotated clinical texts, we have created synthetic FH annotation corpus based on real clinical sentences to promote natural language processing (NLP) tool development [1].
- In the corpus, family members (FM), observation (OBS), age and living status (LIV) were annotated as entities, then all entities related to a family member category are linked into one chain.
- Consequently, the BioCreative/OHNLP 2018 family history extraction task and 2019 NLP Clinical Challenge (N2C2)/OHNLP shared task were enabled [1, 2], that encouraged participants internationally to contribute to FH NLP system development based on clinical narratives.

Objective

 As quality improvement is a continuous process, we aim to further enhance annotation of the corpus in the current study.

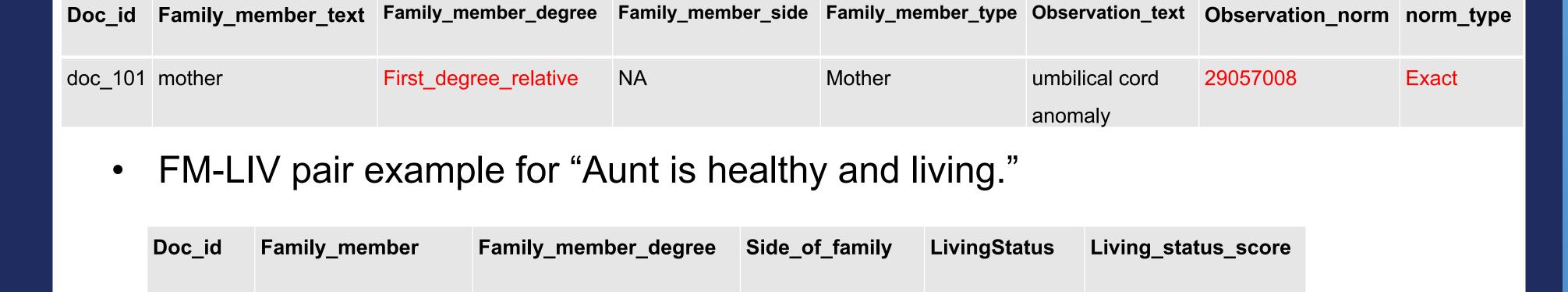
Methods **Entity** normalization ERORR CORECTION Family member -- First degree relative -- Second degree relative training set (2018/2019) -- Third degree relative testing set (2018) testing set (2019) **Observation -- SNOMED code** Adding Removing Correcting Relation **Family member -- Observation** ERORR CORECTION Family member – Living status Resources: One senior annotator + Re-annotation guideline + Automatic transformation of original annotation to Medtator [3] format

Re-Annotation Guideline

- Check if any observation entity was missing from original annotations, if any, we need to add such annotations.
- Cross sentence relation annotation needs to be completed.
- Normalization task is to add SNOMED CT code for each "Observation" in the family history corpus. For normType, it's defaulted as "ExactMatch", For those with no automatic population, manual efforts are needed to fill related SNOMED CT code by looking up https://browser.ihtsdotools.org/ and choose ExactMatch or "ApproximateMatch" as appropriate.
- Given all the new requirements above, all original annotations need to be checked and modified accordingly.

Results and Discussion

• FM-OBS pair example for "His mother has been diagnosed with umbilical cord: anomaly.



• Table 1 shows the statistical comparison between original and enhanced annotations.

Paternal

LivingStatus

- Our exercise demonstrates that the annotation enhancement is feasible when the annotation task is defined clearly.
- The higher-quality synthetic FH annotation corpus would contribute more to the future FH NLP system development.
- https://github.com/OHNLP/fh_eval for more details.

Second degree relative

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References

- 1. Liu, Sijia, Majid Rastegar Mojarad, Yanshan Wang, Liwei Wang, Feichen Shen, Sunyang Fu, and Hongfang Liu. "Overview of the BioCreative/OHNLP 2018 family history extraction task." In *Proceedings of the BioCreative 2018 Workshop*, p. 2018. 2018.
- 2. Shen, Feichen, Sijia Liu, Sunyang Fu, Yanshan Wang, Sam Henry, Ozlem Uzuner, and Hongfang Liu. Family history extraction from synthetic clinical narratives using natural language processing: overview and evaluation of a challenge data set and solutions for the 2019 National NLP Clinical Challenges (n2c2)/Open Health Natural Language Processing (OHNLP) competition. JMIR Medical Informatics 9, no. 1 (2021): e24008.
- 3. He, Huan, Sunyang Fu, Liwei Wang, Sijia Liu, Andrew Wen, and Hongfang Liu. "MedTator: a serverless annotation tool for corpus development." *Bioinformatics* 38, no. 6 (2022): 1776-1778.

Table 1. Statistical comparison between original and enhanced annotations. A: No. span correction, B: No. errors removed, C:

No. newly added, SNO: SNOMED codes, LIV: Living status.

| | | Training set (2018/2019) | | Testing set (2018) | | Testing set (2019) | |
|------------------|--------------|--------------------------|------------------|--------------------|------------------|--------------------|------------------|
| | | Original | Enhanced (A,B,C) | Original | Enhanced (A,B,C) | Original | Enhanced (A,B,C) |
| Document | | 99 | 99 | 50 | 50 | 117 | 117 |
| Chains | | 651 | 761 | 280 | 337 | 631 | 753 |
| Pairs (FM - OBS) | | 754 | 817 | 327 | 355 | 759 | 789 |
| Pairs (FM – LIV) | | 376 | 382 | 161 | 161 | 317 | 381 |
| Age | | 756 | 783 (41,16,43) | 289 | 305 (14,-,16) | 667 | 693 (4,15,41) |
| Living Status | | 415 | 431 (43,1,17) | 181 | 200 (4,-,19) | 391 | 423 (2,10,42) |
| FM | FDR | 802 | 438 (21,4, 12) | 331 | 181 (12,3,2) | 760 | 425 (2,13,12) |
| | SDR | | 331 (35,1,40) | | 144 (12,3,22) | | 320 (-,14, 60) |
| | TDR | | 65 (5,1,10) | | 29 (6,-,5) | | 82 (1, 3, 31) |
| OBS | Entities | 978 | 991 (60,15,28) | 465 | 488 (46,5,28) | 1062 | 1109 (8, 22, 69) |
| | SNO (unique) | - | 1015 (573) | - | 411 (195) | - | 1095 (453) |