

Ped-BERT: Early Detection of Diseases for Pediatric Care Using In Utero Health Information

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Abstract

Artificial intelligence (AI)-based diagnosis systems are particularly relevant in pediatrics given the well-documented impact of early-life health conditions on later-life outcomes. Yet, early identification of diseases for this age group has so far remained uncharacterized, likely because access to relevant health data is severely limited. Thanks to a confidential data use agreement with the California Department of Health Care Access and Information, we are able to develop Ped-BERT: A state-of-the-art deep learning model that accurately predicts the likelihood of 100+ conditions in a pediatric patient’s next medical visit. We link mother-specific pre- and postnatal period health information to pediatric patient hospital discharge and emergency room visits. Our data set comprises 513.9K mother-baby pairs and contains medical diagnosis codes as well as temporal and spatial pediatric patient characteristics, such as age and residency zip code at the time of visit. Following the popular bidirectional encoder representations from the transformers (BERT) approach, we pretrain Ped-BERT via the masked language modeling objective to learn embedding features for the diagnosis codes contained in our data. We then continue to fine-tune our model to accurately predict diagnosis outcomes for a pediatric patient’s next visit, given the history of previous visits and, optionally, the mother’s pre- and postnatal health information. We achieve an area under the receiver operator curve (ROC AUC) of 0.923 and an average precision score (APS) of 0.403. Further, we assess the prediction accuracy of Ped-BERT in identifying a few rare genetic diseases. We also examine its fairness by determining whether prediction errors are evenly distributed across various subgroups of mother-baby demographics and health characteristics, or if certain subgroups exhibit a higher susceptibility to prediction errors.

1 Introduction

2 Early identification of diseases is vital for better treatment options, longer survival rates, improved
3 long-term outcomes, and lower hospital utilization costs. In recent years, breakthrough progress
4 in this area was made by leveraging electronic health records (EHR) and advanced deep learning
5 (DL) architectures, such as convolutional neural networks (CNN, e.g., Nguyen et al. (Deepr)¹),
6 recurrent neural networks (RNN, e.g., Choi et al. (Doctor AI)²), long short-term memory networks
7 (LSTM, e.g., Pham et al. (DeepCare)³), and an even more powerful architecture called bidirectional
8 encoder representation from transformers (BERT). For instance, Li et al.⁴ introduce BEHRT,
9 a BERT-inspired model applied to EHR, capable of predicting the likelihood of more than 300
10 conditions in one's future medical visit; Shang et al.⁵ propose G-BERT, a model that combines
11 the power of graph neural networks (GNN) and BERT for diagnosis prediction and medication
12 recommendation; Rasmy et al.⁶ introduce Med-BERT, also a BERT model, to provide pretrained
13 contextualized embeddings run on large-scale structured EHR.

14 To the best of our knowledge, most advances in this literature (a) rely on EHR representative
15 of the **adult population**;^{7,4} (b) need to specify the patient age distribution;^{1,8,9,2,10,11,6,5} (c) use
16 models that focus on predicting a limited set of health outcomes;^{3,8} (d) focus on improving disease
17 risk assessment performance by accounting only for the timing irregularity between clinical events
18 (e.g., age at the time of visit);^{1,2,4} (e) do not report prediction performance on rare diseases,¹² or
19 (f) do not use in-utero health information for diagnosis prediction.¹²

20 However, computer-aided early detection of diseases holds particular significance in the field of
21 pediatrics. Timely diagnosis and intervention are crucial for enhancing the long-term well-being of
22 children, as highlighted in various studies.^{13,14,15,12} Consequently, we have developed Ped-BERT,
23 an architecture inspired by BERT.¹⁶ Our model accurately predicts over 100 potential diagnoses that
24 a child might face during their upcoming medical appointment. It could serve as a valuable tool for
25 aiding pediatricians in their clinical decision-making processes. Ped-BERT leverages a rich dataset
26 encompassing hospital discharge records and emergency room information for pediatrics, including
27 the patient's age and the residential zip code or county at the time of the visit. Additionally, it

28 can optionally integrate maternal health data from both pre- and postnatal periods. To the best
29 of our knowledge, our prediction framework, leveraging data that matches mother and baby pairs
30 longitudinally is the first of its kind. Furthermore, this dataset empowers us to explore the model's
31 capability to predict rare genetic diseases and to assess its overall fairness, including an examination
32 of whether prediction errors are evenly distributed across different demographics of mother-baby
33 pairs.

34 To summarize, we contribute to the literature as follows: first, we use a novel data set that links
35 medical records of mother-baby pairs between 1991-2017 in California; second, we develop Ped-
36 BERT, a DL architecture for early detection of diseases in pediatric patients seeking care in inpatient
37 or emergency settings; third, we leverage both temporal and spatial patient characteristics, such as
38 age and geographical location at the time of visit; fourth, we also report the model's performance
39 in predicting rare genetic diseases, and fifth, we evaluate Ped-BERT's performance with fairness in
40 mind.

41 Data

42 This study relies on data from the California Department of Health Care Access and Information
43 (HCAI¹⁷). Through a confidential data use agreement, we access the universe of births between 1991
44 and 2012 (Birth data), patient discharge data (PDD), and emergency department visits (EDD)
45 through 2017 from nearly 7,000 California licensed healthcare facilities.¹⁸ We use this data to
46 pre-train and fine-tune Ped-BERT.

47 Birth data

48 We observe over 12M birth records registered in California, including maternal antepartum and
49 postpartum hospital records for the nine months before delivery and one-year post-delivery (Figure
50 1a, top panel). We filter the data to retain only mother-baby pairs (birth IDs) for which the
51 discharge records link to birth certificate data and the baby's social security number (SSN), if the
52 SSN was assigned either at birth or within their first year of life. After filtering, our birth data

53 includes 763,895 mother-baby pairs whose medical records can be tracked over time by linkage with
54 the PDD and EDD data via the SSN (Figure 1b, top panel). Among all variables present in the
55 birth data, we retain information on the baby’s gender, race, and residency zip code and county at
56 birth. We also include information on the mother’s race and education, the month prenatal care
57 began, the number of prenatal visits, and the number of times the mother visited a healthcare
58 facility in an emergency or inpatient setting nine months before and twelve months after birth.

59 **Patient Discharge and Emergency Department Visits**

60 The PDD and EDD datasets consist of over 59M inpatient discharges between 1991 and 2017 and
61 over 81M in emergency visits between 2005 and 2017, respectively (Figure 1a, middle and bottom
62 panels). If the emergency encounter resulted in a same-hospital admission, the inpatient record
63 reflects the emergency encounter, and no separate emergency department visit is recorded.

64 We subset these data to include only those records for which the patient’s SSN has a match in the
65 Birth data (Figure 1b, middle and bottom panels). To improve our machine learning task, we further
66 filter this data to select only those patients whose medical history includes at least three emergency
67 or inpatient stays. After this last filtering, we have nearly 1M inpatient and 2.5M emergency
68 discharge records for 513,963 mother-baby pairs. (Figure 1c, middle, bottom, and top panels). From
69 the PDD and EDD data, we retain information on patient demographic characteristics (including
70 residence zip code and county at the time of visit) and up to three disease codes as listed by the
71 healthcare provider during the encounter. The disease codes in our data are classified using the
72 9th and 10th revisions of the International Statistical Classification of Diseases and Related Health
73 Problems (ICD-9 and ICD-10, respectively). For ease of analysis and interpretability, we convert
74 ICD-10 to ICD-9 codes using the AtlasCUMC dataset^{19,20} and choose to operate at the two-digit
75 sub-chapter level.

76 Via a random split, we use 70% and 30% of these 513,963 mother-baby pairs, respectively,
77 for fine-tuning Ped-BERT and for assessing prediction performance in the downstream task of
78 predicting the next medical diagnosis. In the following, we refer to these two data sets simply as
79 ‘fine-tuning training set’ and ‘fine-tuning test set’.

80 Ped-BERT Pre-training Data

81 For the pre-training of Ped-BERT, it is important to highlight that our goal is to utilize patient
82 records without matches in the fine-tuning data but with available SSN information that enables
83 us to establish connections across time. This distinction is crucial because the data used for pre-
84 training Ped-BERT should not align with our final prediction task to prevent data leakage.

85 We begin with the raw dataset comprising over 59M inpatient discharges (PDD data) and over
86 81M emergency visits (EDD data) (Figure 1a, middle and bottom panels). From this extensive
87 dataset, we retain records of patients with valid SSN. Following this filtering process, we are left
88 with nearly 3.8M inpatient stays and 16.2M emergency visits, corresponding to nearly 5.5M patient
89 IDs (Figure 1d). Subsequently, we exclude all patients whose SSN match the 513,963 birth IDs
90 described in the previous subsection because we will use this data for fine-tuning Ped-BERT (Figure
91 1e). Finally, to improve our machine learning task, we further refine the data to include only patients
92 with a minimum of three medical encounters. This step leaves us with approximately 2M inpatient
93 discharges and 10M in emergency room visits, totaling 1,855,013 unique patients for pre-training
94 Ped-BERT (Figure 1f).

95 Via a random split, we use 80% and 20% of these data, respectively, for pre-training Ped-BERT
96 and testing prediction performance. In the following, we refer to these two data sets simply as
97 ‘pre-training training set’ and ‘pre-training test set’.

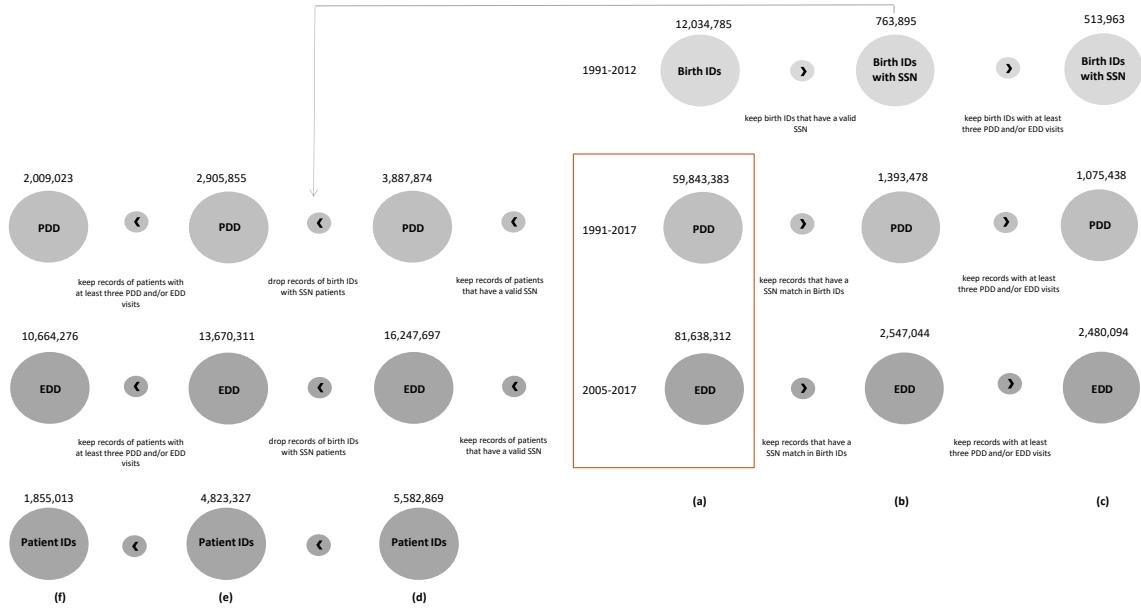


Figure 1: Filtering, linking, and summary of our data. (a-b) From the initial set of 12M birth IDs, 59.8M patient discharge data records (PDD), and 81.6M emergency department data records (EDD), we only retain those that can be linked via SSN at birth or in the first year of life: 764K, 1.4M and 2.5M, respectively. (c) We further filter by number of inpatient/emergency encounters, only retaining records for patients with at least three medical encounters. This final set consists of approximately 3.5M hospital visits (PDD and EDD combined) between 1991-2017 for 513,963 mother-baby pairs. This data is used for fine-tuning Ped-BERT. (d) From the initial set of 59.8M PDD records and 81.6M EDD records, we only retrain those that can be linked via SSN at some point in life: 2.9M and 13.6M, respectively. (e-f) We further drop the records of patients whose SSN has a match in the 513,963 mother-baby pairs data or have less than three inpatient/emergency encounters. This final set consists of around 2M and 10M records in the PDD and EDD data, respectively, corresponding to 1,855,013 unique patient IDs. We use this data for pre-training Ped-BERT.

98 Patient Medical History

99 For our fine-tuning task of predicting diagnosis in the upcoming medical visit, we rely on
100 patient health information, starting nine months before birth, until data censoring. Let \mathbf{P}
101 represent our sample of patients, and \mathbf{T} represent a set of sorted time stamps. In our
102 data, each patient $p \in \{1, 2, \dots, P\}$, is described by a set of birth attributes, $p.A_b =$
103 $\{A_1, A_2, \dots, A_n\}$ recorded in the prenatal period and/or at the time of birth. Each pa-

tient is also characterized by a set of inpatient/emergency encounter attributes, $p.A_e = \{(A_1, A_2, \dots, A_n|1), (A_1, A_2, \dots, A_n|2), \dots, (A_1, A_2, \dots, A_n|T)\}$ recorded at time $t \in \{1, 2, \dots, T\}$ of encounter with the medical provider. The attributes in $p.A_b$ cover the baby's gender and race, mother's race and education, pregnancy month prenatal care began, the number of prenatal visits, mother inpatient/emergency visits nine months before and twelve months after birth, and residency zip code/county at birth. Similarly, the attributes in $p.A_e$ are sequences of patient disease codes, patient age, and patient residency zip code/county at the time of visit. Figure 2a illustrates, in tabular form, the medical history of a hypothetical patient with birth attributes (data column 2) $p.A_b = \{\text{female, hisp, hisp, < high school, 2, 9, 1, 3, 94002}\}$ and medical encounter attributes (data columns 3-7) $p.A_e = \{ ([D1, D2], 0, 94002 | \text{visit}=1), ([D1], 4, 94002 | \text{visit}=2), \dots, ([D1], 7, 91000 | \text{visit} = 5)\}$. The diagnosis codes assigned by medical personnel are represented as D1, D2, ... etc.

Descriptive statistics of the data utilized for fine-tuning Ped-BERT are presented in Figure 2. The distribution of the baby/patient's race is approximately even between males and females; both the baby/patient's and mother's race are predominantly white or Hispanic/other; most mothers have attained an educational level below high school or have completed college; prenatal care typically starts within 1-3 months of conception, with most mothers receiving 10-12 prenatal care visits; a majority of mothers in our data did not require inpatient or emergency room services in the prepartum and postpartum period (see Figure 2b for additional details). Lastly, mother-baby pairs in our data are well-distributed across California (Figure 2c).



Figure 2: Patient medical history and descriptive statistics. (a) Example, in tabular form, of a patient’s medical history documenting data collected in the in-utero period or at the time of birth, and during the first five inpatient/emergency visits. (b-c) Summary statistics for mother-baby/patient demographics and health-related outcomes belonging to the 513,963 mother-baby pairs used for fine-tuning of Ped-BERT. **Abbreviations:** F = Female, M = Male, AS.PI = Assian_Pacific Islander, Bl = Black, Hisp_Oth = Hispanic_Other, NAm_EA = Native, Am_Eskimo_Aleut, Wh = White, <HS = less than High-school, grad = graduate education, b. = before, a. = after, unkn = unknown.

¹²³ Methods

¹²⁴ This study aims to introduce Ped-BERT, a BERT transformer-encoder-based architecture.^{21,16}
¹²⁵ Ped-BERT consists of a bidirectional training procedure and masked language modeling approach
¹²⁶ (MLM), which enable the model to learn the probability distribution of different diagnosis outcomes
¹²⁷ in a pediatric patient’s next inpatient or emergency visit. We describe our methodology below.

¹²⁸ Models

¹²⁹ We decompose our prediction task into two components. In the first step, we pre-train our Ped-
¹³⁰ BERT model using each patient’s health attributes data, $p.A_e$, and BERT’s MLM approach. The
¹³¹ objective here is to learn good disease representations. Afterward, via the second step, we fine-tune
¹³² Ped-BERT’s parameters in a supervised fashion via the downstream task of predicting the diagnosis
¹³³ in the next medical visit.

¹³⁴ Ped-BERT Pre-training

¹³⁵ The pre-training stage is concerned with learning good disease embeddings. Concretely, Ped-BERT
¹³⁶ pretrains bidirectional diagnosis representations from medical histories by jointly conditioning both
¹³⁷ left and right diseases in a pediatric patient’s medical history. This approach has been shown to
¹³⁸ outperform other deep learning architectures, such as CNN, RNN, and LSTM,^{1,2,7,3} or left-to-right
¹³⁹ attention as presented in the original transformer architecture.²¹ In addition, Ped-BERT is pre-
¹⁴⁰ trained using the MLM approach, whose objective is to randomly replace a fraction of the diagnosis
¹⁴¹ codes with mask tokens [MASK] and task the model with predicting these hidden disease codes
¹⁴² instead.

¹⁴³ This stage relies on the unlabeled pre-training data split into ‘pre-training training set’ and
¹⁴⁴ ‘pre-training test set’, and for simplicity, Figure 3a illustrates the pretraining task of Ped-BERT
¹⁴⁵ using as an example the hypothetical patient introduced earlier (see Figure 2a). First, the model is
¹⁴⁶ given the patient’s health history in the following format: [CLS] $D_1 D_2$ [SEP] D_1 [SEP] $D_1 D_2$
¹⁴⁷ [SEP] D_1 [SEP] D_1 [SEP]. Here, [CLS] is a token denoting the beginning of the patient’s medical

148 history, and the [SEP] token is added to indicate the end of a medical visit. Both tokens, [CLS]
149 and [SEP], are added to aid with the subsequent diagnosis prediction task. The D tokens represent
150 up to three medical diagnoses at the time of visit (see Figure 3a - Patient Diagnosis History)

151 Second, the data undergoes pre-processing for the MLM task, involving the random selection
152 of 15 percent of the disease tokens for masking (see Figure 3a - Masking). The selection/masking
153 process follows the original BERT model.¹⁶

154 Third, a trainable input embedding matrix is created. We first identify the unique diagnosis
155 codes in the masked training data, map them to integer values, and then encode each patient’s
156 diagnosis history using this mapping. Since our disease sequences have different lengths, we use
157 zero padding as a placeholder for adjusting sequence length. We continue by encoding information
158 on visit position, patient’s age and geographical location to give our model a sense of the timing,
159 age, and location of events. While age embeddings have been used before (e.g., BEHRT⁴), the
160 geographical location is unique to Ped-BERT. We hypothesize that one’s location could be an
161 essential determinant of health outcomes due to the environmental impacts of the quality of local
162 resources, such as clean air and safe water, for example. These resources are prerequisites for
163 health, and poor attributes can be particularly detrimental to vulnerable populations such as the
164 very young. We pre-train Ped-BERT using the ‘pre-training training set’ with different input
165 embedding specifications. We define our baseline specification as the sum of diagnosis and positional
166 embeddings. We then assess for any MLM prediction performance improvement by adding age and
167 location embeddings (see Figure 3a - Embeddings).

168 Finally, the output of the input embeddings sublayer is sent to multi-head attention and feed-
169 forward network sublayers (see Figure 3a - transformer-encoder stack). The multi-head attention
170 sublayer is followed by post-layer dropout and normalization. The output is passed to the fully
171 connected feedforward network sublayer and followed by post-layer normalization. This last layer
172 produces the logits for each token in the diagnosis vocabulary. The predicted masked token is
173 extracted from these logits using a Softmax activation function, which provides a probability dis-
174 tribution over each diagnosis token in the vocabulary (see Figure 3a - MLM Predictions). We keep
175 the ‘pre-training test set’ for final model evaluation (see Figure 3a - Evaluation).

176 Ped-BERT Fine-tuning for Diagnosis Prediction

177 A complete training procedure of Ped-BERT includes fine-tuning the model for specific downstream
178 tasks using labeled data. Our main task in the fine-tuning stage is to predict the probability
179 distribution over a set of diagnosis codes in a pediatric patient’s next inpatient or emergency room
180 visit. Figure 3b shows the workflow for applying the pre-trained Ped-BERT to this predictive task.

181 We start from the labeled fine-tuning data split into ‘fine-tuning training set’ and ‘fine-tuning
182 test set’. For each patient p and each data partition, we randomly choose a visit index v ($2 \leq$
183 $v < T$) to split their health attributes data, $p.A_e$ into input-output pairs. The input is denoted
184 by $X_{p.A_e} = \{(A_{\text{disease codes}}, A_{\text{age}}, A_{\text{zip}}|1), \dots, (A_{\text{disease codes}}, A_{\text{age}}, A_{\text{zip}}|v)\}$ and the output by $y_{p.A_e}$,
185 which is a multi-hot vector of length 105 (corresponding to the total number of disease codes in Ped-
186 BERT’s vocabulary) equal to 1 for diagnosis codes that exist in the next visit, $A_{\text{disease codes}}|v + 1$.
187 We tokenize and encode the diagnosis history of each patient, and feed the data into Ped-BERT for
188 embeddings extraction (based on the output of the last layer of the transformer-encoder block, see
189 Figure 3b - Preprocessing). We then use the ‘fine-tuning training set’ to fine-tune all Ped-BERT’s
190 learned parameters by fitting and optimizing a multiclass logistic regression model for subsequent
191 diagnosis prediction (see Figure 3b - Learning). We keep the ‘fine-tuning test set’ until the very
192 end for the final model evaluation (see Figure 3b - Evaluation).

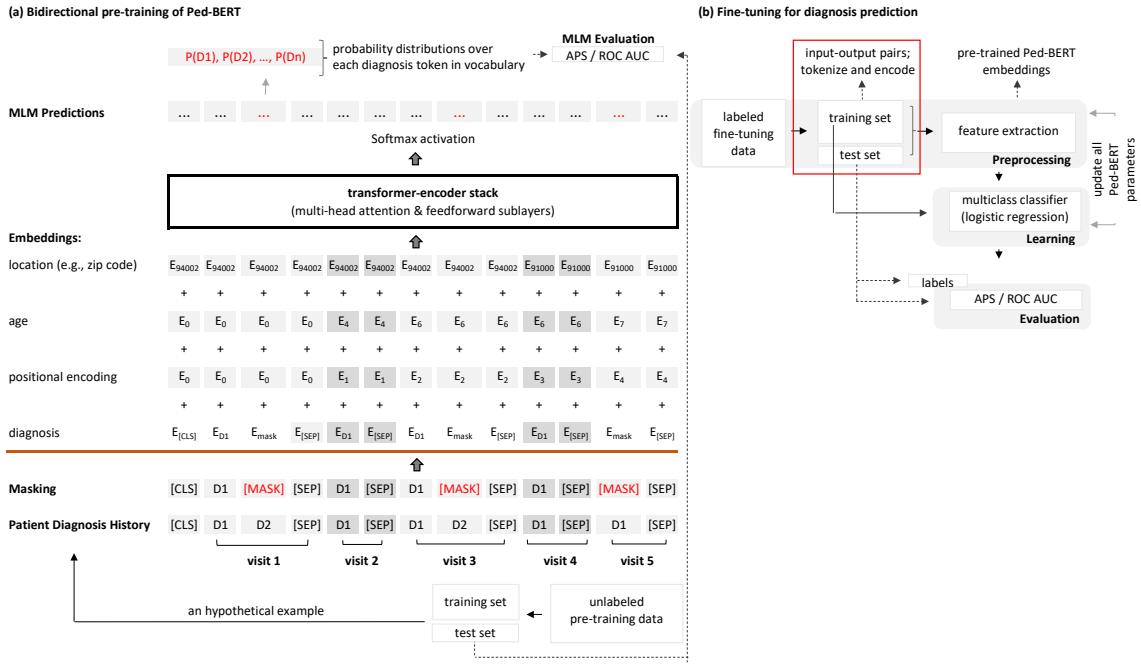


Figure 3: **Ped-BERT architecture.** (a) The pre-training task is explained using as an example the hypothetical patient introduced in Figure 2a: Ped-BERT sees the medical history and masks some of the diagnosis codes before sending them to embedding, multi-head attention, and feed-forward network sublayers. The task here is to predict the [MASK] disease codes. (b) In the fine-tuning task, the pre-trained Ped-BERT model parameters are fine-tuned using a logistic model with the objective of predicting the probability distribution over given diagnosis codes in a pediatric patient’s next inpatient or emergency room visit. (a-b) The fine-tuning and pre-training steps are evaluated using the APS and ROC AUC scores.

193 Prediction Performance Evaluation

194 We evaluate the performance of both the pre-trained and fine-tuned Ped-BERT model for disease
 195 prediction using two key metrics: the Average Precision Score (APS) and the Area Under the
 196 Receiver Operating Curve (ROC AUC). Note that the APS summarizes a precision-recall curve as
 197 the weighted mean of precisions achieved at each threshold, with the increase in recall from the
 198 previous threshold used as the weight (see scikit-learn²² for implementation details).

199 During the pre-training phase, we calculate these metrics by comparing the model’s predictions
 200 to the actual ground-truth data associated with the [MASK] token for all patients within the ”pre-

201 training test set.” In the fine-tuning stage, we represent the model’s predictions for each patient p
202 as $y_{p.A_e}^*$ and gauge the model’s performance by assessing the agreement between these predictions
203 ($y_{p.A_e}^*$) and the actual values ($y_{p.A_e}$). This assessment is conducted by computing the APS and
204 ROC AUC on the ”fine-tuning test set” individually for each patient, and subsequently calculating
205 the averages across all patients for all diagnosis codes, as well as the averages across all patients for
206 each specific diagnosis code.

207 Results

208 We present results from Ped-BERT’s pre-training stage and then evaluate Ped-BERT’s fine-tuned
209 ability to predict the diagnosis in the subsequent medical encounter for all disease codes, and
210 separately, for rare genetic conditions. We conclude by discussing the results of a few fairness tasks
211 and how Ped-BERT could guide medical practitioners.

212 Ped-BERT Pre-training Evaluation

213 The optimal architecture of Ped-BERT has the following specifications: the input diagnosis embed-
214 ding matrix is of size 120 x 128, with the first dimension representing the length of the diagnosis
215 vocabulary (115 unique two-digit diagnosis codes + *OOV* + [MASK] + [CLS] + [SEP] + padding
216 token) and the second dimension representing the embedding size; the patient history is restricted
217 to a maximum length of 40 tokens; the encoder is a stack of 6 identical layers; inside each of these
218 identical layers there is a multi-head attention sublayer containing 12 heads and a feedforward net-
219 work sublayer containing 128 hidden units; dropout regularization rate is set to 0.1; pre-training is
220 for 15 epochs using the Adam optimizer with a learning rate of $3e - 5$ and a decay of 0.01.

221 Ped-BERT is pre-trained using different specifications for the input embedding matrix. As
222 mentioned in the Methods section, we define our baseline embeddings specification as the sum
223 of diagnosis embeddings and positional encodings. We then augment this baseline by adding age
224 embeddings (+ age), zip embeddings (+ zip), county embeddings (+ cnty), age + zip embeddings
225 (+ age + zip), and age + county embeddings (+ age + cnty). Figure 4a presents a couple of

interesting findings derived from the ‘pre-training test set’: adding age embeddings slightly improves the APS score relative to baseline [0.52 vs. 0.51]; adding county embeddings to the baseline + age specification results in negligible APS differences [APS: 0.521 vs. 0.52]; adding additional embeddings (such as age and/or county) to the baseline specification results in negligible differences in terms of ROC AUC. We also assess specifications with the patient’s zip code instead of the county given as additional embeddings and find that the model performance is below the base specification in terms of both APS and ROC AUC (results not presented in Figure 4a). In summary, our results suggest that, in the context of pediatric patients, augmenting a pre-trained model with information on the patient’s age at the time of medical encounter has a modest positive impact on model performance, while the addition of patient’s county of residence at the time of the visit does not improve the results. For more information on the distributional details regarding the data used to pre-train Ped-BERT see Supplementary Figure S1).

We proceed to evaluate the quality of our pre-trained embeddings through both intrinsic and extrinsic methods. Intrinsic assessment involves examining the embeddings’ quality through visual inspection and reporting cosine similarity among disease embeddings. For the extrinsic evaluation, we examine the embeddings’ effectiveness in predicting patient gender distribution for specific disease codes.

To visually inspect Ped-BERT’s embeddings, we reduce the embedding space to 2D using t-SNE (see scikit-learn²³ for implementation details). Figure 4b shows the reduced embeddings for the baseline + age input embeddings specification. The visualization reveals that similar diseases (such as those related to injury and poisoning, diseases of the respiratory system, and birth conditions) cluster together. Furthermore, diseases known to frequently co-occur (such as neoplasms, diseases of the blood, and blood-forming organs) are also grouped closely. Upon closer examination of these 2D disease embedding clusters, a remarkable association with the International Classification of Disease Codes (ICD codes) becomes evident. Notably, this finding is interesting because we did not explicitly provide this information to Ped-BERT during the pre-training phase. Subsequently, we proceed to report the cosine similarity between disease codes using Ped-BERT’s learned embeddings. Upon aggregation at the chapter level, we observe a range of similarity values, with the minimum

²⁵⁴ and maximum values being -0.318 and 1, respectively; the values at the 25, 50, and 95 percentiles,
²⁵⁵ are 0.093, 0.229, and 0.586, respectively (additional details are available in supplementary Figure
²⁵⁶ S2).

²⁵⁷ Finally, we conduct an extrinsic evaluation of Ped-BERT's embeddings by assessing their perfor-
²⁵⁸ mance in predicting the gender distribution of patients with congenital anomalies and tuberculosis.
²⁵⁹ This evaluation is prompted by the increasing body of evidence highlighting sex-specific disparities
²⁶⁰ in the prevalence of congenital anomalies and tuberculosis, with research studies demonstrating
²⁶¹ higher prevalence rates among pediatric males.^{24,25} As shown in Supplementary Figure S3, Ped-
²⁶² BERT consistently predicts a higher prevalence of these two diseases among males when evaluated
²⁶³ on the 'pre-training test set', with a Fisher's exact test value equal to 0.0862 ($p < 0.1$).

²⁶⁴ In summary, our current intrinsic and extrinsic evaluation results indicate that Ped-BERT has
²⁶⁵ developed a substantial understanding of the contextual relationships between diseases.

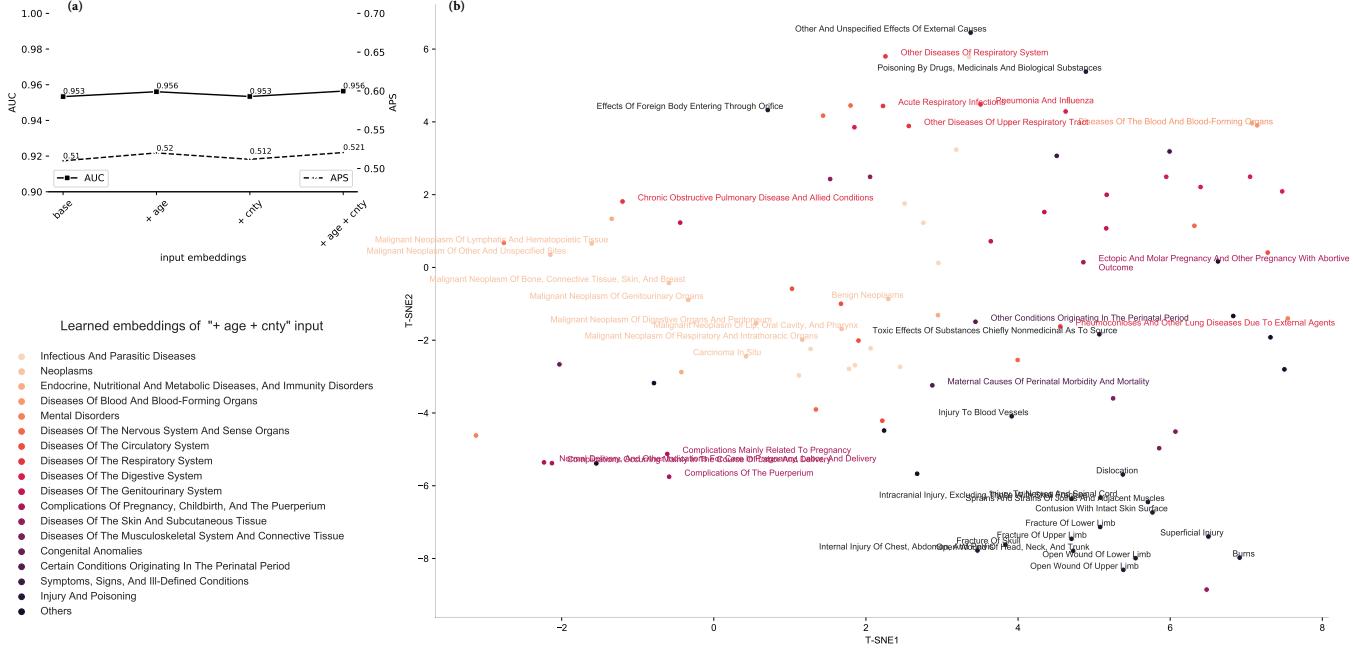


Figure 4: Evaluation of Ped-BERT’s MLM task. (a) The average precision score (APS, right y-axis) and the area under the receiver operating curve (ROC AUC, left y-axis) were computed as sample averages for the following embedding specifications: base (which is the sum of diagnosis embeddings and positional encodings), base + age, base + county, and base + age + county embeddings. These metrics represent comparisons between the ground truth (unmasked tokens) and the MLM-predicted diagnosis (masked tokens) in the test data. (b) Intrinsic evaluation of the MLM embeddings via visual inspection for the base + age input embeddings specification. We reduce the dimension of the embedding matrix from 120×128 to 120×2 using t-SNE to create a 2D visualization of all 115 two-digit diagnosis codes in our vocabulary. Colors represent diagnosis chapters.

266 Ped-BERT Fine-tuning for Diagnosis Prediction

267 A complete training procedure of Ped-BERT includes fine-tuning. Ped-BERT is not designed for
 268 any specific task in the pre-training step but instead trained as a general disease model for pediatric
 269 patients. In the fine-tuning stage, we generalize Ped-BERT to predict the medical diagnosis in the
 270 subsequent inpatient or emergency pediatric visit. Specifically, we update the pre-trained model
 271 parameters for our specific downstream task using regular supervised logistic learning on labeled
 272 data by adding on top of the pre-trained Ped-BERT a feedforward layer with 64 hidden units and

273 an output layer containing a softmax activation function. The model is trained for 100 epochs using
274 the Adam optimizer with a learning rate of $3e - 4$ and early stopping. In Figure 5a (black lines),
275 we focus on reporting results related to the base embeddings specification and its corresponding
276 augmentations with age (+ age) and age + county (+ age + cnty). We find no differences in
277 ROC AUC (continuous black lines) and very small differences in APS (dashed black lines) across the
278 three pre-trained embedding specifications (e.g., APS base: 0.392, base + age: 0.397, base + age
279 + cnty: 0.399). For comparison, BEHRT’s⁴ performance in the downstream task of predicting the
280 subsequent diagnosis codes for the adult population, shows a one-point difference in APS and an
281 inisgnificant difference in ROC AUC between the base and base + age embeddings.

282 In Figure 5b, we report the ROC AUC for each diagnosis code in our ‘fine-tuning test set’ as
283 derived from the base + age embeddings specification; we highlight the top five (blue colors) and the
284 least five performances (red colors) in terms of AUC scores. Our results indicate that Ped-BERT
285 exhibits high predictive performance for certain conditions, including maternal causes of perinatal
286 morbidity and mortality (AUC = 0.983), malignant neoplasm of genitourinary organs (AUC =
287 0.950), congenital anomalies (AUC = 0.938), ischemic heart disease (AUC = 0.918), malignant
288 neoplasm of bone (AUC = 0.906), and organic psychotic conditions (AUC = 0.901). On the other
289 hand, it demonstrates lower prediction performance for conditions like injury of nerves of spinal
290 cord (AUC = 0.619), malignant neoplasm of respiratory and intrathoracic organs (AUC = 0.615),
291 toxic effects of substances (AUC = 0.614), persons with potential health hazards related to personal
292 and family history (AUC = 0.531), and other spirochetal diseases (AUC = 0.410).

293 In Figure 5c, our focus centers on assessing the suitability of Ped-BERT for detecting rare
294 genetic diseases for pediatric patients. To achieve this, we compute and report the ROC AUC
295 scores for various genetic diseases, including other Diseases of the biliary tract (AUC = 0.645),
296 other metabolic and immunity disorders (AUC = 0.598), diseases of white blood cells (AUC =
297 0.649), cerebral degenerations manifesting in childhood (AUC = 0.656), congenital anomalies of
298 eyes (AUC = 0.895), and diseases of the capillaries (AUC = 0.588). These results indicate varying
299 levels of prediction performance for these rare diseases, ranging from decent to suboptimal. For
300 more details, please refer to Supplementary Table S2, which provides additional information on the

301 number of patients with these rare diseases in both the ‘fine-tuning training set’ and ‘fine-tuning
302 test set’.

303 **The Role of Ped-BERT Pre-training and Birth Attributes Data**

304 To further explore the efficacy of Ped-BERT’s pre-training and the role of birth attributes data, we
305 conduct two additional investigations. First, we compare the performance of a disease prediction
306 model using randomly initialized base embeddings against the three models employing pre-trained
307 Ped-BERT embeddings. In Figure 5a (black lines), we observe a significant enhancement in APS
308 and a modest improvement in ROC AUC performance when comparing the model with randomly
309 initialized base embeddings to the one with pre-trained base Ped-BERT embeddings (APS: 0.372
310 vs. 0.392, ROC AUC: 0.915 vs. 0.92). Second, we extend the analysis by incorporating birth
311 attributes data ($p.A_b$) into both the randomly initialized and pre-trained Ped-BERT embedding
312 models to assess potential improvements in disease prediction. In Figure 5a (maroon lines), notable
313 distinctions are only evident for the model with randomly initialized base embeddings (APS: 0.372
314 vs. 0.392, ROC AUC: 0.915 vs. 0.922) and the model with pre-trained base Ped-BERT embeddings
315 (APS: 0.392 vs. 0.403, ROC AUC: 0.92 vs. 0.926). These results suggest that a pre-trained
316 Ped-BERT model with base embeddings is a good substitute for a model with randomly initialize
317 embeddings that also require birth attributes data for better prediction performance.

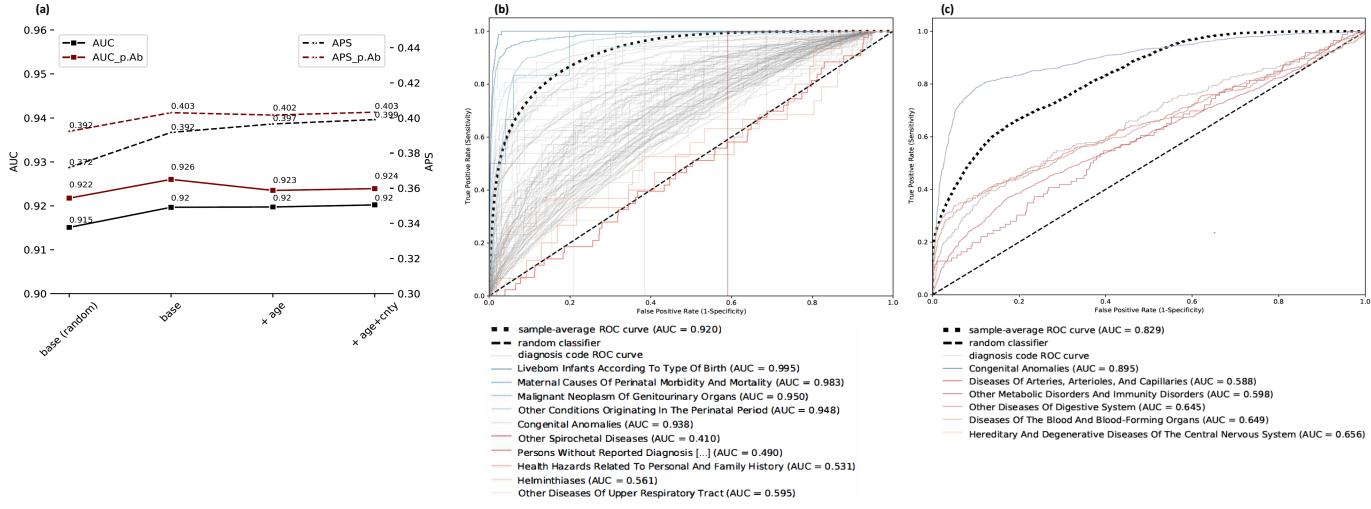


Figure 5: Evaluation of the Disease Prediction task. We consider the following embedding specifications: black lines = randomly initialized embeddings (base (random)), pre-trained base Ped-BERT embeddings, pre-trained base + age Ped-BERT embeddings, and pre-trained base + age + county Ped-BERT embeddings; maroon lines = augment the four models with features from the birth attributes data (*p.Ab*). **(a)** The APS (right y-axis) and ROC AUC (left y-axis) computed for each embedding specification scenario outlined above (black lines and maroon lines)). These metrics represent comparisons between the ground truth and the predicted diagnosis for each patient in the output partition of the input-output pairs of our test dataset. **(b)** True Positive Rates and False Positive Rates curves averaged across all patients for each diagnosis in the data (grey lines), for the top five (blue lines) and least five (red lines) diagnosis codes based on AUC scores, and average across all patients for all diagnosis codes in the data (dot-dashed black lines); the long-dashed line denotes a random classifier. **(c)** similar to **(b)** but for selected rare genetic diseases for top one (blue lines) and least five (red lines).

318 Fairness Tasks

319 We are interested in determining whether next-visit diagnosis prediction errors are uniform across
 320 subgroups in our data. Figure 5 already gives us some insights into the model’s APS and ROC
 321 AUC performance (overall and by disease code), but it is desirable to understand how well it
 322 performs for different subgroups. For example, Figure 2 identifies groups of mother-baby/patient
 323 demographics and health-related outcomes belonging to the pairs used in this analysis. Our data
 324 also contains information on the mother’s country of birth, which is rarely available to research
 325 and unique to our study. As such, in this section, we aim to assess the fine-tuned Ped-BERT’s

³²⁶ prediction performance with fairness in mind and use the ‘fine-tuning test set’ and the pre-trained
³²⁷ baseline + age embeddings for this task.

³²⁸ We find minimal differences in ROC AUC performance across groups of patient gender and
³²⁹ race, mother race and education, month prenatal care began, the number of prenatal visits, and the
³³⁰ number of times the mother visited a healthcare facility overnight or in an emergency setting (see
³³¹ Figure 6, top and middle panels). Next, we create bins for the mother’s country at her own birth,
³³² for similar patient ages, for zip codes/counties belonging to the same geographical region,²⁶ and for
³³³ similar PM 2.5 pollution values at the time of birth.²⁷ We find that Ped-BERT is more susceptible
³³⁴ to prediction errors depending on the mother country of origin at her own birth, for patients in the
³³⁵ age subgroups 3-17 and greater than 17 ((AUC: 0.933 and 0.901) than those in the 0-2 subgroup
³³⁶ (AUC: 0.871), and for patients that have been born in a zip code with unhealthy pollution (AUC:
³³⁷ 0.887) as opposed to moderate or good pollution (AUC: 0.907 and 0.914, respectively)(see Figure
³³⁸ 6, bottom panel).

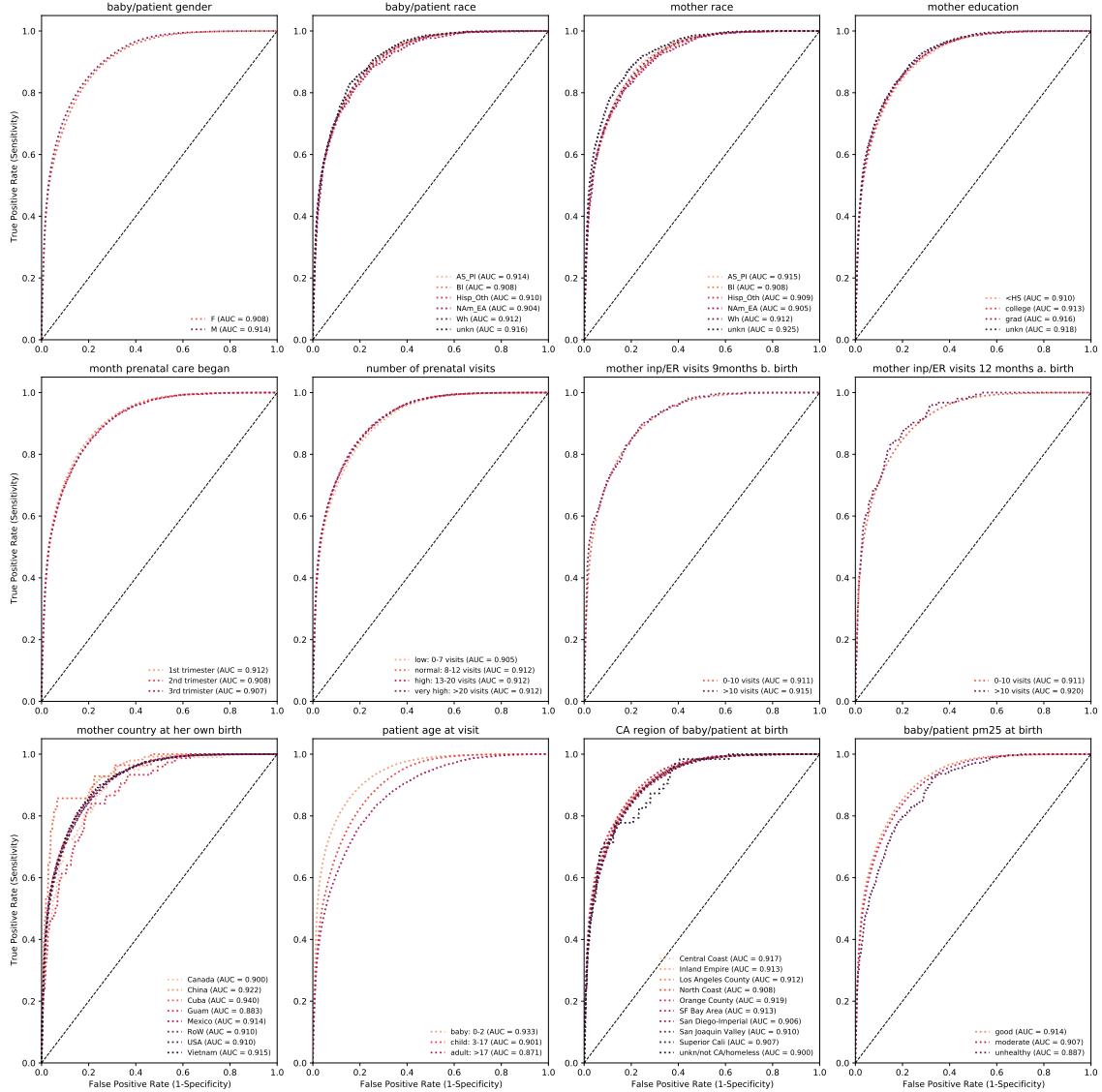


Figure 6: Fairness tasks: Using results from the fine-tuning stage, we compare evaluation results across different subgroups (e.g., baby/patient gender and race; mother race and education; month prenatal care began). The evaluation results rely on the fine-tuned model with base + age embeddings applied to the test sample. True Positive Rates (Sensitivity) vs. False Positive Rates (1-Specificity) are shown as red dots lines. A long-dashed line denotes a random classifier.

339 **Model Application**

340 Our results suggest that Ped-BERT may provide useful information to clinical providers. We
341 imagine the approach can be utilized in two ways. First, a medical provider can use the fine-tuned
342 Ped-BERT model with base + age embeddings to augment clinical decision-making with machine
343 learning. Using our model's predictions would reduce uncertainty over the most likely conditions.
344 The table below provides a prediction example for a patient randomly chosen from our 'fine-tuning
345 test set'. The model is presented with the patient's previous two-digit ICD9 health history (along
346 with age information at the time of visit - not presented here for simplicity). The model outputs
347 a probability distribution over all diagnosis codes in Ped-BERT's fine-tuning vocabulary. Listed
348 below are the top five predicted diseases.

User: Medical provider
Previous two-digit ICD9 health history

[CLS] 46 78 [SEP] 48 [SEP] 49 48 [SEP] 46 [SEP] 07 [SEP]
TOP five diagnosis predictions at the next medical encounter

ICD9 code: 49 48 46 78 38
probability: 0.255 0.160 0.153 0.098 0.033
two-digit ICD9 code descriptions

07: Infectious And Parasitic Diseases
38: Diseases Of The Ear And Mastoid Process
46: Acute Respiratory Infections
48: Pneumonia And Influenza
49: Chronic Obstructive Pulmonary Disease And Allied Conditions
78: Symptoms

349 A second way that a medical professional could use our approach would be to fine-tune the pre-
350 trained Ped-BERT on their own corpus of medical records and make predictions for new patients.

351 **Discussion**

352 This research aims to improve the early detection of diseases in pediatric patients using a unique
353 healthcare database and the latest developments in bidirectional encoder representations from trans-

354 formers (BERT). The data used in our analysis consists of vital statistics and birth information,
355 as well as hospital discharge data and emergency room visits in California between 1991 and 2017
356 for 513,963 mother-baby pairs. A BERT-based model called Ped-BERT is trained using a masked
357 language model (MLM) approach and is able to accurately predict the likelihood of over 100 condi-
358 tions in a child’s next medical visit. The study also evaluates Ped-BERT’s prediction performance
359 for rare genetic disorders, and for fairness by assessing whether prediction errors are uniformly
360 distributed across different mother-baby demographics and health characteristics subgroups. The
361 model has the potential to assist clinical providers in making machine learning-augmented decisions
362 about pediatric healthcare.

363 The pre-training stage of Ped-BERT involves learning good representations of diseases by testing
364 different combinations of input embeddings to represent a patient’s health history. The baseline
365 specification is the sum of diagnosis embeddings and positional encodings. Age and zop/county
366 embeddings augment this baseline in our performance improvement tests. We find that adding
367 age embeddings improves the APS score relative to baseline, and further expanding with county
368 embeddings results in negligible APS differences relative to the baseline + age specification. We
369 use intrinsic and extrinsic methods to evaluate the embedding quality further. Intrinsically, we find
370 that the model has learned to cluster together diseases that belong to the same ICD chapter or
371 are known to co-occur. Extrinsicly, we find that the disease embeddings generated by Ped-BERT
372 correctly predict the male-skewed gender distributions for congenital anomalies and tuberculosis.

373 The fine-tuning stage of the Ped-BERT model involves adapting the model for the specific
374 downstream task of predicting the diagnosis in the subsequent inpatient or emergency pediatric
375 visit. The results averaged across all patients and disease codes show insignificant differences in
376 ROC AUC and minor differences in APS across the baseline, baseline + age, and baseline + age
377 + cnty fine-tuned embedding specifications. The ROC AUC sample averages were also computed
378 across all patients and (a) each diagnosis code, highlighting the top five and least five performances,
379 and (b) six rare genetic diseases.

380 Finally, we assess the fine-tuned Ped-BERT for fairness, as models that perform poorly on
381 certain subgroups can lead to unequal outcomes and perpetuate biases. In this case, Ped-BERT

³⁸² performs generally well, with some differences based on the mother's country at her own birth, the
³⁸³ patient's age, and patient's pollution exposure at birth.

³⁸⁴ We propose several possible directions for future research based on the architecture and prop-
³⁸⁵ erties of Ped-BERT. Its ability to encode diagnosis codes, age, and geographical location into a
³⁸⁶ fixed-length vector representation can make it useful for many tasks. For example, one can focus on
³⁸⁷ fine-tuning Ped-BERT for early detection of rare genetic pediatric conditions. It is also worth noting
³⁸⁸ here that the specific dataset used can significantly impact the model's performance. For example,
³⁸⁹ Ped-BERT was pre-trained on a dataset of medical records from California between 1991-2017, so
³⁹⁰ it may not perform as well on tasks that involve other states in the US or other countries. Another
³⁹¹ possibility is increasing the training size for older patients and those living in less environmentally
³⁹² friendly areas. The rationale here is that a more diverse training set will expose the model to a
³⁹³ broader range of ages and geographical locations by making the location embeddings more powerful
³⁹⁴ for learning good disease representations while helping the model generalize better to new tasks.

³⁹⁵ Availability of Data and Code

³⁹⁶ We collected health data from The California Department of Health Care Access and Information
³⁹⁷ (HCAI¹⁷), which provides confidential patient-level data sets to researchers eligible through the
³⁹⁸ Information Practices Act (CA Civil Code Section 1798 et seq.). Note that researchers interested
³⁹⁹ in working with this health data should request it directly from HCAI (<https://hcai.ca.gov/data-and-reports/research-data-request-information/>) as it is HIPAA protected, and by
⁴⁰⁰ agreement, we are not allowed to distribute it.
⁴⁰¹

⁴⁰² The geospatial data comes from the Census Bureau and includes 2010 ZCTA shapefiles
⁴⁰³ (https://www.census.gov/cgi-bin/geo/shapefiles/index.php?year=2010&layergroup=ZIP+Code+Tabulation+Areas_),²⁸ 2010 county shapefiles (<https://www.census.gov/cgi-bin/geo/shapefiles/index.php?year=2010&layergroup=Counties+%28and+equivalent%29>),²⁹
⁴⁰⁴ 2010 ZCTA to county codes (<https://www.census.gov/programs-surveys/geography/technical-documentation/records-layout/2010-zcta-record-layout.html>),³⁰ ZCTA to
⁴⁰⁵

408 zip codes crosswalks (https://github.com/censusreporter/acs-aggregate/blob/master/crosswalks/zip_to_zcta/ZIP_ZCTA_README.md),³¹ as well as the 2020 geographical division of
409 California's 58 counties into ten regions (<https://census.ca.gov/regions/>).²⁶
410

411 The PM 2.5 pollution data is made available by the Atmospheric Composition Analysis
412 Group of the Washington University of St. Luis (<https://sites.wustl.edu/acag/datasets/historical-pm2-5-across-north-america/>).³² It provides information on concentrations of am-
413 bient fine particulate matter across North America, which combines data from chemical transport
414 modeling, satellite remote sensing, and ground-based monitoring.
415

416 The underlying code for this study is publicly available at https://github.com/corneliailin/CA_hospitals_online.
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- 500

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⁵⁰² **Declaration of interests:** The authors declare no competing interests.

503 **Appendices**

504 **A Data Acquisition and Processing**

505 **A.1 Health Data**

506 Our data is distributed by the California Department of Health Care Access and Information
507 (HCAI¹⁷). We requested the following files for research purposes:

508 *Linked Birth Files (Birth data)*: a research database created to study delivery and birth out-
509 comes. It includes maternal antepartum and postpartum hospital records for the nine months before
510 delivery and one-year post-delivery. In addition, the linked file contains birth records and all infant
511 readmissions occurring within the first year of life. The file contains all infants born in a given year,
512 including births that happened in a California hospital that reported to HCAI, births that occurred
513 in a California hospital that did not report to HCAI, and births that occurred outside California.
514 It includes all infants and mothers, irrespective of whether they were linked to a birth record. The
515 linked pairs of birth/delivery records have information associated with a mother/baby pair from
516 the baby's discharge data record, the mother's discharge data record, and the birth certificate data.
517 Linked birth files are available beginning with the 1991 calendar year reporting period (HCAI¹⁷).

518 *The Patient Discharge Dataset (PDD)*: consists of a record for each inpatient discharge from
519 a California-licensed hospital. Licensed hospitals include general acute care, acute psychiatric,
520 chemical dependency recovery, and psychiatric health facilities. These datasets are available starting
521 in 1983 (HCAI¹⁷). For more information on the data and reporting requirements, see the California
522 Inpatient Data Reporting Manual.³³

523 *The Emergency Department Dataset (EDD)*: includes information from hospitals licensed to
524 provide emergency medical services. The EDD encounters include those patients who had face-
525 to-face contact with the provider. If the patient left without being seen, the patient would not
526 have had a face-to-face encounter with a provider, and therefore the EDD encounter would not be
527 reported. These data sets are available beginning January 2005 (HCAI¹⁷).

528 Our study's primary variable of interest is the primary, secondary, and tertiary ICD 9 or ICD10
529 diagnosis codes at the time of visits. We accessed it along with other relevant metadata, such as
530 mother-baby demographics and mother health-related outcomes nine months before and 12 months
531 after birth.

532 A.2 Geospatial Data

533 The geospatial data was constructed and made available by the Census Bureau. For California,
534 the relevant 2010 ZCTA and county-specific shapefiles,^{28,29} the 2010 ZCTA to county codes,³⁰ and
535 the ZCTA to ZIP crosswalks³¹ were identified and mapped to our health data for visualization and
536 analysis purposes. For the Fairness analysis, we extracted the California - Census 2020 geographical
537 division of counties into regions. Table S1 contains a summary of the counties used for each region.

Table S1: **Geographical division of California's counties** - Details of counties included in each California region to support the Fairness analysis presented in Figure 6.

Region	County
Central Coast	Monterey, San Benito, San Luis Obispo, Santa Barbara, Santa Cruz, Ventura
Inland Empire	Riverside, San Bernardino
Los Angeles County	Los Angeles
North Coast	Del Norte, Humboldt, Lake, Mendocino, Napa, Sonoma, Trinity
Orange County	Orange
SF Bay Area	Alameda, Contra Costa, Marin, San Francisco, San Mateo, Santa Clara, Solano
San Diego - Imperial	Imperial, San Diego
San Joaquin Valley	Alpine, Amador, Calaveras, Madera, Mariposa, Merced, Mono, San Joaquin, Stanislaus, Tuolumne, Fresno, Inyo, Kern, Kings, Tulare
Superior Cali	Butte, Colusa, El Dorado, Glenn, Lassen, Modoc, Nevada, Placer, Plumas, Sacramento, Shasta, Sierra, Siskiyou, Sutter, Tehama, Yolo, Yuba

538 A.3 PM2.5 Pollution Data

539 PM2.5 pollution data comes from the Atmospheric Composition Analysis Group³² of the Washington
540 University of St. Luis. According to the source, this data is the estimated concentrations of
541 ambient fine particulate matter across North America, which combines information from chemical
542 transport modeling, satellite remote sensing, and ground-based monitoring. The estimates included
543 information from updated historical emissions inventories and meteorological data, fine resolution
544 satellite-based estimates of PM2.5, and ground-based measurements of PM2.5, PM10, and total sus-

545 pended particles (TSP) measurements. We extracted information at the monthly level for each zip
 546 code in our data to construct groups according to the EPA definition for healthy ($0.0\text{--}12.0\mu\text{g}/\text{m}^3$),
 547 moderate ($12.1\text{--}35\mu\text{g}/\text{m}^3$), and unhealthy ($> 35\mu\text{g}/\text{m}^3$) PM2.5 pollution exposure at birth.

548 **B Supplementary Figures**

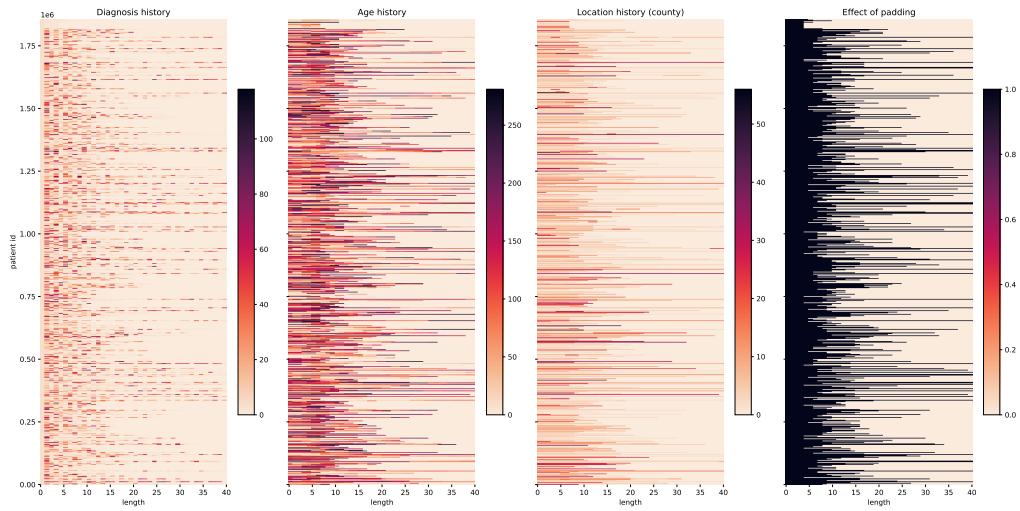


Figure S1: **Summary statistics of encoded input for pre-training Ped-BERT.** The x-axis represents the length of a given patient history, which we optimally set to 40 periods. Each tick on the y-axis represents a diagnosis, age, location history, and padding summary for a given patient ID in the pre-training data. Heatmap values and colors represent: for (a-c), the encoded disease codes, age, and location history; for (b): the effect of zero padding since not all patients have a history length equal to 40.

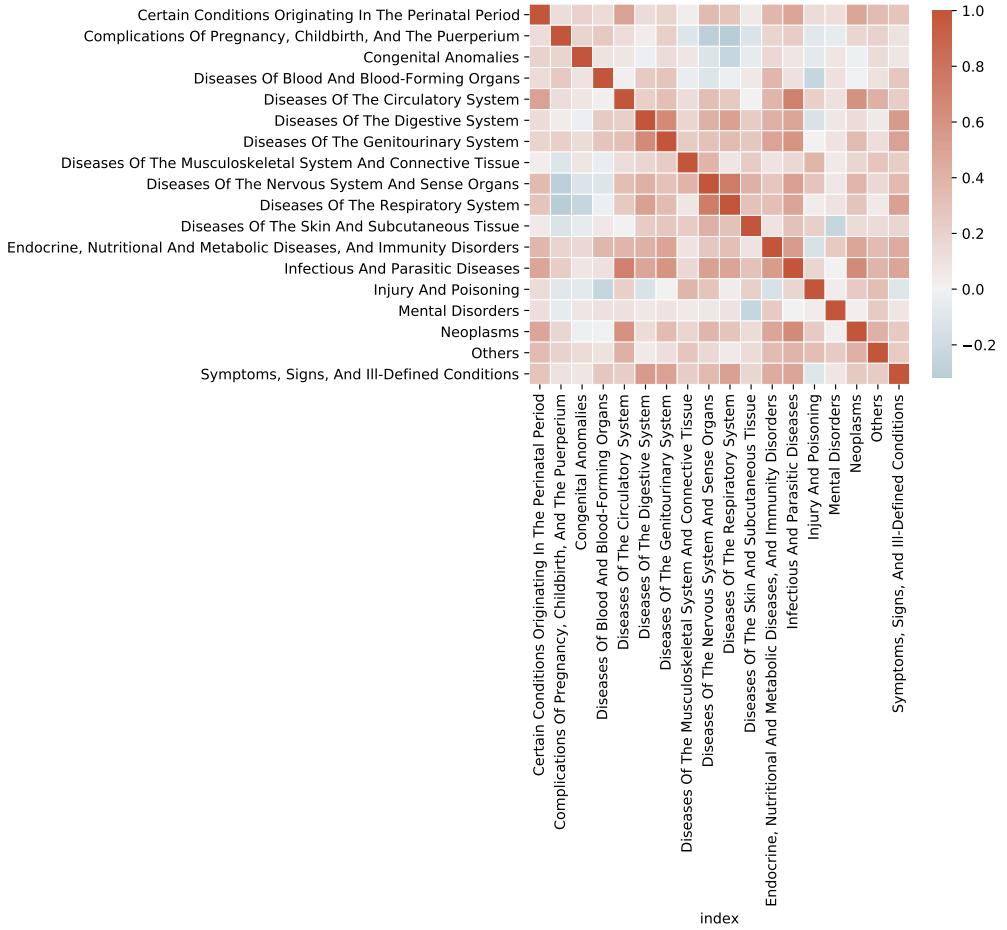


Figure S2: Intrinsic evaluation of embeddings. Learned embeddings are extracted from the pre-training stage for the base + age input embedding specification. The heatmap represents the cosine similarity for all the diagnosis codes in our data aggregated at the chapter level. Negative values (blue shades) reflect opposite similarities, and positive values (red shades) represent close similarities.

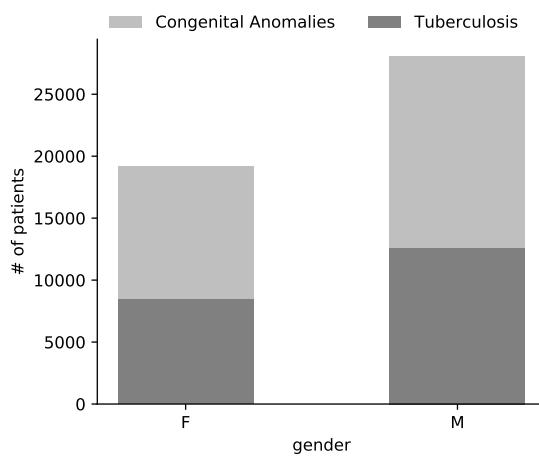


Figure S3: **Extrinsic evaluation of embeddings.** Assess the performance of the pre-trained Ped-BERT model in predicting the patient gender distribution for congenital anomalies (light gray) and tuberculosis (dark gray). The results presented here rely on the base + age embeddings specification using the ‘pre-training test set’. Abbreviations: F = Female, M = Male.

Table S2: **Rare Genetic Diseases Prediction.** Number of patients in the ‘fine-tuning training set’ and ‘fine-tuning test set’ for selected rare genetic diseases at the two-digit ICD9 code level.

two-digit ICD9 diag code	# of patients in training data	# of patients in test data
27	3620	1541
28	1186	480
33	447	183
44	178	82
57	799	333
74	3816	1641