Reliable Learning of Bernoulli Mixture Models

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Abstract In this paper, we have derived a set of sufficient conditions for reliable clustering of data produced by Bernoulli Mixture Models (BMM), when the number of clusters is unknown. A BMM refers to a random binary vector whose components are independent Bernoulli trials with cluster-specific frequencies. The problem of clustering BMM data arises in many real-world applications, most notably in population genetics where researchers aim at inferring the population structure from multilocus genotype data. Our findings stipulate a minimum dataset size and a minimum number of Bernoulli trials (or genotyped loci) per sample, such that the existence of a clustering algorithm with a sufficient accuracy is guaranteed. Moreover, the mathematical intuitions and tools behind our work can help researchers in designing more effective and theoretically-plausible heuristic methods for similar problems.

1 Introduction

Demixing data samples from mixture models, also called model-based clustering, has long been studied by statisticians and computer scientists. Although, plenty of promising algorithms have been introduced in this area, c.f. [1, 2, 3, 4], fewer efforts have been focused on deriving theoretical guarantees on reliable clustering of data samples. The aim of this paper is to elaborate on this issue, i.e. deriving analytical guarantees on clustering accuracy, for a particular case of interest: Bernoulli Mixture Models (BMM).

A Bernoulli model refers to a random binary vector \( \mathbf{X} = [x_1, \ldots, x_L] \in \{0, 1\}^L \) with independent components, where \( x_i \) is Bernoulli trial with probability of success \( p_i \), i.e. \( x_i \sim \text{Bern}(p_i) \). Let us denote the frequency vector as \( \mathbf{\hat{p}} \triangleq [p_1, \ldots, p_L] \in [0, 1]^L \). Let \( f(\mathbf{X}; \mathbf{\hat{p}}) \) denote a Bernoulli model with frequency vector \( \mathbf{\hat{p}} \):

\[
f(\mathbf{X}; \mathbf{\hat{p}}) \triangleq \prod_{\ell=1}^{L} p_\ell^{X_\ell} (1 - p_\ell)^{1-X_\ell}.
\]

A BMM is a mixture of a finite number of Bernoulli models [5]. Mathematically, it can be expressed as

\[
\mathbb{P}\{\mathbf{X}\} = \sum_{k=1}^{K} w_k f\left(\mathbf{X}; \mathbf{p}^{(k)}\right),
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\mathbb{P}\{\mathbf{X}\} = \sum_{k=1}^{K} w_k f\left(\mathbf{X}; \mathbf{p}^{(k)}\right),
\]
where $K$ denotes the number of clusters (or mixture components), $\mathbf{p} = \{\mathbf{p}^{(1)}, \ldots, \mathbf{p}^{(K)}\}$ are the latent frequency vectors associated to the clusters, and $\mathbf{w} = \{w_1, \ldots, w_K\}$ are cluster probabilities with $\sum_k w_k = 1$. In our statistical inference scenario, $K$, $\mathbf{p}$ and $\mathbf{w}$ are all assumed to be hidden.

The above mathematical setting has exact analogies in population genetics studies [6], where researchers aim to cluster individuals based on their multilocus genotype data, i.e. binary sequences of a given length indicating presence or absence of certain variants in genome. In genetics literature, the frequency vector is denoted by the vector of allele frequencies, while each dimension of $\mathbf{X}$ indicates a genotyped locus. In the absence of familial relationships among individuals, which is the case in the majority of current researches, individuals and thus their genotype sequences can be assumed to be independently distributed from each other [7, 8]. Moreover, by choosing non-neighboring genetic variants, statistical independence among different variants of the same sequence exists almost surely [9, 10, 7]. Variations among allele frequencies of different populations are usually due to different genetic ancestries or environmental factors correlated with geographical habitats of individuals [7]. In this paper, the notions from the mathematical view of BMMs, and those frequently used by computational geneticists have been used interchangeably.

Basic concepts of BMMs have a long history in statistics [11, 12, 13, 14]. However, it was initially used by geneticists in [6], a seminal paper in population genetics, where authors proposed an MCMC-based algorithm for the inference of population structure by clustering and parameter estimation in an iterative manner. In recent years, several statistical frameworks have been introduced by researchers for the same purpose, with different performances on real and synthetic datasets. However, fundamental limits for reliable clustering or parameter estimation in a BMM, in terms of minimum required dataset sizes or sequence lengths have not been derived. Also, sufficient conditions under which “good” approximations would be algorithmically possible have remained a mystery. To the best of our knowledge, this paper is the first mathematically rigorous study toward this goal, i.e. deriving a set of sufficient conditions under which reliable inference of genetic population structure from multi-locus genotyped data is possible.

1.1 Our Contribution

We will look at clustering the dataset into a finite number of partitions such that the majority of samples from each partition belong to a single component of the underlying BMM. Let us denote such partitions qualitatively as pure clusters. We have proposed a measure called Mutual Dependence Measure (MDM) to quantify the pureness of any given cluster, based on the following fundamental difference between a Bernoulli model and a BMM. In a Bernoulli model different dimensions are statistically independent\(^1\), contrasting their dependency in a BMM\(^2\). This implies that the mutual information between any two dimensions is zero in a Bernoulli model, while there exist pairs of components with non-zero mutual information in a BMM.

MDM is a generalized version of the mutual information which can be defined among more than

\(^1\)According to the genetic counterpart of BMMs, this statement holds as long as variants are not in vicinity of each other in genome.

\(^2\)Statistical dependence between pairs of Bernoulli trials in BMM with $K \geq 2$ has already been mentioned in the literature; See [13] for more details. However, quantifying the level of this dependency through information-theoretic measures has been initially carried out in this paper.
two random variables. We show that the empirical values of MDM on a subset of the dataset becomes significantly large if the subset includes samples from different mixture components. However, it would be negligible for sufficiently pure subsets, where samples are more likely to be drawn from only one mixture component. This analytical tool is used as a criterion to accept/reject any given partitioning of the dataset.

Using MDM, we derive a set of conditions under which a sufficiently correct learning of BMMs is possible. Mathematically, consider a dataset of \( n \) i.i.d. binary vectors, each of dimension \( L \), which are drawn from an unknown BMM. The number of clusters underlying the model is also assumed to be unknown, making the problem setting more suitable in practical situations. Assume at least \( \mathcal{L} \) out of the \( L \) Bernoulli trials are informative, i.e. their success rate (frequency) differ between different mixtures. We have shown that if \( \mathcal{L} > \mathcal{L}_{\min} \), and \( n \geq n_{\min}(\mathcal{L}, L) \), then there exists a clustering algorithm whose output on the dataset resembles the true mixtures components of the BMM with high probability. Here, \( \mathcal{L}_{\min} \) and \( n_{\min} \) are both functions of BMM parameters.

1.2 Related Works

This paper, viewed from a mathematical side, relates to the fundamentals in inference of BMMs. From an application side, however, it is related to a well-studied problem in population genetics, i.e. clustering individuals based on genotype data. Accordingly, we briefly review a number of studies related to both areas.

Employment of BMMs in order to model multi-dimensional categorical data goes back to [16], while more detailed mathematical and historical explanations can be found, for example, in [15, 17, 18]. In two classic works [14] and [19], a series of heuristic measures have been introduced to assess the number of mixture components in a BMM; However, their performance is validated only through experimental investigations. Authors in these papers have conjectured that learnability is possible as long as independence holds between cluster parameters, while their studies lack a rigorous sufficiency analysis. Also, fundamental limits in the inference of the number of clusters are not derived. From an algorithmic point of view, Expectation-Maximization (EM) is the most widely used framework for statistical inference in BMMs; See [20] and [18]. In [21], a popular EM-based technique for unsupervised learning of finite mixture models (including BMMs) is introduced, which makes no assumption on the number of mixture components. Also, see [22] for another well-cited paper on model-based clustering of mixture model data. In [23], EM is employed to tune proper initializations for existing inference algorithms on BMMs. From a theoretical perspective, a set of statistical guarantees on the convergence of EM algorithm in mixture model problems has been recently given in [24], however, authors have mainly focused on Gaussian distributions rather than Bernoulli models.

Our work is also related to Bayesian non-parametric approaches in the sense that the number of clusters is open-ended, and will be inferred based on the observed data. Some good reviews on non-parametric approaches in statistics can be found, for example, in [25, 26, 27]. In particular, [28] has proposed a unified non-parametric framework for model-based clustering with the use of hierarchical Dirichlet mixtures. Almost all of the studies reviewed so far share a common property: at their best, authors have only proved convergence to a sub-optimal likelihood value, rather than providing guarantees on the accuracy of the final clustering/learning.
From a geneticist point of view, this paper basically builds upon the statistical model presented in [6]. Although the algorithm proposed in [6] (STRUCTURE) basically makes use of a more general framework, i.e. binomial mixture models, however its applications mostly fall into the realm of BMMs since majority of genetic variants are biallelic [9]. A large number of studies since then indicate the biological plausibility of using BMM for population genetics studies of unrelated individuals. More discussions on the biological aspects of this model can be found, for example, in [8], [29] and [30]. In [31], authors have performed a simulation study based on [6], in order to assess the number of clusters in a given population. A number of recent and/or popular softwares for computational population analysis can be found in [30], [32], [33] and [34], which mostly focus on binary datasets, the same configuration of data we have considered in this paper. A generalization of [6], which also takes population admixtures into account, is presented in [35]. The problem setting which is tackled by the current work encompasses both models described in [6] and [35], since we have made no restrictive assumptions, such as independence, on latent allele frequencies. The role of population stratification in genome-wide association studies (GWAS), an important application of genetics research, are discussed in [8] and [36]. For more research on the employment of BMMs for correcting GWA studies, see [37], [38], [39] and [40].

The paper is organized as follows: Section 2 introduces mathematical notation and definitions that are used throughout the paper. In Section 3.1, the basic idea mentioned above is discussed with mathematical details, where Section 3.2 proposes a more practically suitable form of our results. In Section 4 we derive a set of sufficient conditions and a computationally exhaustive algorithm which is shown to achieve a reliable clustering of data with high probability. Finally, conclusions are made in Section 5.

2 Notation and Definitions

We define \( B \) as a BMM with the following specifications: \( K \) clusters (or mixture components), set of frequency vectors \( \{ p^{(1)}, \ldots, p^{(K)} \} \), and cluster probabilities \( w = \{ w_1, \ldots, w_K \} \). Let us denote the number of Bernoulli trials (loci) per sample by \( L \). In this regard, we denote \( p = \left[ p^{(1)} | \ldots | p^{(K)} \right]^T \in [0,1]^{K \times L} \) as the frequency matrix associated to \( B \).

Assume \( X_1, \ldots, X_n \in \{0,1\}^L \) to be \( n \) i.i.d. samples (or sequences) drawn from \( B \). For the ease of notation, we define \( X \) both as the set \( \{X_1, \ldots, X_n\} \) and the matrix \( [X_1 | \ldots | X_n]^T \in \{0,1\}^{n \times L} \) representing the collection of samples. A clustering (or alternatively partitioning) of data samples in \( X \), denoted by \( Z \in \{1,2,\ldots\}^n \), is an integer vector where \( Z_i \) represents the cluster index to which data sample \( X_i \) is assigned to. In this regard, we denote \( J \in \{1,2,\ldots,K\}^n \) as the latent true clustering of data samples. The following definitions are used multiple times in this paper:

**Definition 1.** An \( \epsilon \)-pure subset of sequences in \( X \), is a subset where a fraction of more than \( 1 - \epsilon \) of sequences are drawn from one mixture component of the BMM, according to \( J \).

Accordingly, one can define an \( \epsilon \)-correct clustering of \( X \) as follows:

**Definition 2.** An \( \epsilon \)-correct clustering of \( X \) is a partition \( Z \in \{1,2,\ldots\}^n \) where all clusters are \( \epsilon \)-pure with respect to \( J \).
For the mixture model to be theoretically learnable, different mixture components may not have exactly the same set of parameters. For example, in the case of a BMM, it is crucial to assume that different mixture components possess sufficiently different frequency vectors. In this regard, we need to formally define a measure of deviation among allele frequency vectors \( \{ p^{(1)}, \ldots, p^{(K)} \} \), which will be helpful in later sections.

**Definition 3.** An allele frequency matrix \( p \in [0,1]^{K \times L} \) is \((L, \delta)\)-separable, if for each pair of rows of \( p \) there exist at least \( L \leq L \) loci where the difference between frequencies of each location exceeds \( \delta \).

Finally, a clustering algorithm \( \psi \) is a mapping from the set of data samples to a partitioning with an arbitrary number of clusters, i.e. \( \psi : \{0,1\}^{n \times L} \rightarrow \{1,2,\ldots\}^{n} \).

## 3 Reliable Inference: Results and Ideas

The following theorem provides a set of non-asymptotic conditions on \( n \) and \( L \), for which an almost correct clustering of data generated by a BMM with unknown parameters is algorithmically possible.

**Theorem 1.** Assume \( \mathcal{B} \) to be a BMM with \( K \) clusters, frequency matrix \( p \in [0,1]^{K \times L} \) and cluster probabilities \( w = \{w_1, \ldots, w_K\} \), where \( K \), \( p \) and \( w \) are latent. \( p \) is assumed to be \((L, \delta)\)-separable for some \( L \leq L \) and \( \delta > 0 \). Let \( X = \{X_1, X_2, \ldots, X_n\} \) be a set of \( n \) i.i.d. samples drawn from \( \mathcal{B} \). Also, assume there exists \( 0 < \alpha \leq 1 \) such that the smallest cluster in \( X \) has at least \( \alpha n \) samples. Let \( \epsilon > 0 \) and \( \zeta > 0 \) be such that

\[
L > L_{\min} \triangleq \frac{4L^*72L^* \log K}{\alpha \epsilon^2 (1 + \log \frac{1}{\alpha \epsilon})^2}
\]

and

\[
n \geq L_{\min} \log K \max \left\{ \frac{1}{L^* \epsilon^3 \log \frac{4eKL2L^*}{\zeta L^*}}, \frac{1 + \log \frac{2K}{L^*}}{1 - L_{\min}/L^*} \right\},
\]

with \( L^* \triangleq \frac{1 - \alpha}{2\alpha(1 - \epsilon)^2} (1 + \log \frac{1}{\alpha \epsilon}) \). Then, there exists a clustering algorithm \( \psi : \{0,1\}^{n \times L} \rightarrow \mathbb{N}^n \), such that \( \psi(X) \) is \( \epsilon \)-correct on \( X \) with probability at least \( 1 - \zeta \).

Proof of Theorem 1 together with the algorithm \( \psi \) which achieves the claimed accuracy are given in Section 4. Before that, Sections 3.1 and 3.2 extensively discuss the core mathematical idea behind this results.

### 3.1 Mutual Dependence Measure

Assume a random sequence \( R \in \{0,1\}^L \) whose distribution corresponds to a BMM with specifications described in Section 1 and a set of parameters \((K, w, p)\). If we condition on \( R \) being drawn from a particular mixture component, for example \( k \in \{1,2,\ldots,K\} \), then the statistical distribution of \( R \) would be

\[
\mathbb{P} \{ R|k \} = \prod_{\ell=1}^{L} \mathbb{P} \{ R_{\ell}|k \}.
\]

(2)
However, the general distribution of $R$ without this assumption is $\mathbb{P}\{R\} = \sum_k w_k \mathbb{P}\{R|k\}$, as shown in \(1\). A more subtle look at $P$ and $Q$ simply reveals that in a mixture model, unlike the case of a single Bernoulli model, different loci are not independent from each other. This argument can be qualitatively justified as follows: a group of loci can convey information about the origin of the whole sequence $R$, which then impacts the distribution of any other group of loci. However, this flow of information does not occur when sequence is known to be generated from a single Bernoulli model, e.g. $\mathbb{P}\{R|k\}$.

Based on the above argument, we can introduce the following Mutual Dependence Measure (MDM) in order to quantify whether a group of sequences are more likely to be drawn from a single model, or a mixture of various models with different parameters.

**Definition 4.** Assume a binary matrix $Q \in \{0,1\}^{n \times L}$. The Mutual Dependence Measure (MDM) of $Q$, denoted by $D(Q)$, is defined as

$$D(Q) \triangleq \frac{1}{n} \sum_{i=1}^{n} \log \left( \frac{\{ \ell | Q_j = Q_i \} / n}{\prod_{j=1}^{L} \hat{p}_\ell (1 - \hat{p}_\ell)^{1 - Q_j,\ell}} \right),$$

where $Q_i$ denotes the $i$th row of $Q$, $\{\cdot\}$ denotes the cardinality of a set, and $\hat{p}_\ell$ is defined as follows:

$$\hat{p}_\ell \triangleq \frac{1}{n} \sum_{i=1}^{n} Q_{i,\ell}, \quad \ell = 1, \ldots, L,$$

which indicates the empirical frequency of the $\ell$th column in $Q$.

In other words, $D(Q)$ is the Kullback-Leibler divergence between two distributions $P_1$ and $P_2$, where: $P_1$ is the actual empirical distribution of the rows in $Q$, while $P_2$ denotes a distribution of rows computed as the product of empirical marginals of separate entries. Note that we have $D(Q) = D_{KL}(P_1 || P_2) \geq 0$.

Assume rows of $Q$ to be $n$ i.i.d. samples drawn from a single Bernoulli model with an arbitrary frequency vector, i.e. $K = 1$. Then, based on the law of large numbers and also \(2\), it can be readily seen that

$$\lim_{n \to \infty} D(Q) = D_{KL}\left( \lim_{n \to \infty} P_1 || \lim_{n \to \infty} P_2 \right) = 0.$$

However, the following lemma which also forms the mathematical core of this paper, shows that $D(Q)$ deviates from zero with high probability whenever $Q$ is generated by a BMM with $K \geq 2$ mixture components. This property can be used as a tool for determining whether a given subset of sequences in $X$ is $\epsilon$-pure or not.

**Lemma 1.** Assume $\mathbb{B}$ to be a BMM with $K$ clusters, $L$ Bernoulli trials per sample, frequency matrix of $p$ and cluster probabilities of $\phi = \{\phi_1, \phi_2, \ldots, \phi_K\}$ with $\sum_k \phi_k = 1$. Let $p$ to be $(\mathcal{L}, \delta)$-separable for some $\mathcal{L} \leq L$ and $\delta > 0$. Also, assume $\phi_k \leq 1 - \epsilon$, for all $k = 1, 2, \ldots, K$ and some $\epsilon > 0$. Consider $Y_1, \ldots, Y_n$ to be $n$ i.i.d. samples drawn from $\mathbb{B}$, and let $Y = [Y_1| \ldots | Y_n]^T$. Then, if $\mathcal{L} > \frac{L \epsilon}{K-1} \triangleq \frac{1 + \log \frac{\epsilon}{\delta^2}}{2(1-\epsilon) \delta^2}$, we have

$$\mathbb{P}\{D(Y) \leq \tau\} \leq 2^{L+1} e^{-\beta n},$$

where $\tau \triangleq \epsilon (1 - \epsilon) \delta^2 \left( \mathcal{L} - \frac{L \epsilon}{K-1} \right) > 0$ and $\beta \triangleq \frac{\tau^2}{L \epsilon^2}$. 

6
Proof of Lemma 1 is given in Appendix A. The assumption of \( \phi_k \leq 1 - \epsilon \) for all \( k \), yields that for \( n \to \infty \), the set of observations \( Y_1, \ldots, Y_n \) will not be \( \epsilon \)-pure almost surely. Hence, for an asymptotically large non-\( \epsilon \)-pure set of observations, we have \( D(Y) \gtrsim \tau \).

On the other hand, we have already discussed that for a completely pure set, i.e. when samples are drawn from a single Bernoulli model or equivalently for \( K = 1 \), we have \( \lim_{n \to \infty} D(Y) = 0 \). The following lemma will provide tail-bounds on the deviation of \( D(Y) \) when \( Y \) consists of a finite number of samples drawn from a single Bernoulli model.

**Lemma 2.** Assume \( \tilde{B} \) to be a single Bernoulli model with \( L \) Bernoulli trials per samples and an arbitrary frequency vector. Consider \( Y_1, \ldots, Y_n \) to be \( n \) i.i.d. samples drawn from \( \tilde{B} \), and let \( Y = [Y_1|\ldots|Y_n]^T \). Then, we have
\[
P\{D(Y) \geq \tau\} \leq 2L^L + 1 e^{-\beta L^2 n},
\]
where \( \tau \) and \( \beta \) are the same as in Lemma 1.

The proof for Lemma 2 is also given in Appendix A.

### 3.2 Maximal Mutual Dependencies

According to Lemmas 1 and 2, it can be readily seen that the number of samples required to make reliable assessments grows exponentially with respect to \( L \), i.e. \( n \gg L^L / \tau^2 \). This fact should not be surprising since reliable computation of \( D(\cdot) \) is subject to having a relatively close estimation of an \( L \)-dimensional binary distribution. Hence large values of \( L \) are both theoretically and practically unsuitable for estimating \( D(Y) \). To overcome this, one may rely only on a small subset of loci for decision making which is not statistically reliable as it does not exploit all the information embedded in the data. A better alternative, however, is to compute numerous mutual dependencies over various subsets of loci, and then combining the results; For example by taking their maximal value.

**Definition 5.** The maximal Mutual Dependence Measure (m-MDM) of a binary matrix \( Q \in \{0,1\}^{n \times L} \) is defined as
\[
D_{\text{max}} (Q; L_0) \triangleq \max_{S \subseteq \text{col}(Q), |S| = L_0} D(S),
\]
where \( L_0 \leq L \), and \( \text{col}(Q) \) denotes the set of all columns of \( Q \). Maximization is taken over all \( \binom{L}{L_0} \) possible sub-matrices of \( Q \) with size \( n \times L_0 \).

\( D_{\text{max}} \) will be very helpful when the number of informative trials \( L \) is significantly larger than \( L_0 \). Using this modified measure, the following theorem shows that the error rate in detecting impure subsets of samples drops exponentially with respect to both \( n \) and \( L \).

**Theorem 2.** Assume \( \tilde{B} \) to be a BMM with \( K \) clusters, \( L \) Bernoulli trials per sample, frequency matrix of \( p \) and cluster probabilities of \( \phi = \{\phi_1, \ldots, \phi_K\} \) with \( \sum_k \phi_k = 1 \). Consider \( Y_1, \ldots, Y_n \) to be \( n \) i.i.d. samples drawn from \( \tilde{B} \) and let \( Y = [Y_1|\ldots|Y_n]^T \).

1) For \( K \geq 2 \), assume \( p \) is \((L, \delta)\)-separable for some \( \mathcal{L} \leq L \) and \( \delta > 0 \). Also, assume \( \phi_k \leq 1 - \epsilon \)
for some $\epsilon > 0$ and all $k$. Assume $L > L_c \triangleq \frac{K-1}{2(1-\epsilon)\delta^2} \left(1 + \log \frac{K}{\epsilon}\right)$. For some $L_0$ with $L_c < L_0 \leq L$, let $\tau \triangleq \left(1-\epsilon\right)\delta^2 \left(\frac{1}{K-1} - \frac{1}{K}\right) \left(L_0 - L_c\right)$. Then, we have

\[
P\{D_{\text{max}}(Y; L_0) \leq \tau\} \leq 2^L \left(\frac{L}{L_0}\right)^2 \exp \left(\frac{-\tau^2 n L}{L_0^2 2^{L_0+1}}\right).
\]

2) For $K = 1$ and any $\tau > 0$ we have

\[
P\{D_{\text{max}}(Y; L_0) \geq \tau\} \leq \left(\frac{L}{L_0}\right)^2 2^{L_0+1} \exp \left(\frac{-\tau^2 n L}{L_0^2 2^{L_0+1}}\right).
\]

Given that $L \gg \frac{K-1}{2(1-\epsilon)\delta^2} \left(1 + \log \frac{K}{\epsilon}\right)$ and $n \gg f(K, L, \epsilon, \delta)$ with $f$ being a corresponding function, we can see that probability of misdetection between a pure (or “good”) subset of sequences with a “bad” one is strictly bounded. In fact, Theorem 2 provides us with a mathematically rigorous and reliable criterion for distinction among correct and incorrect partitionings of data samples in finite databases. The next section will present a corresponding clustering algorithm together with supporting mathematical analysis for reliable inference of population structures, i.e. clustering of data samples and estimating cluster parameters.

4 Reliable Clustering: Algorithm and Analysis

This section will present a simple and computationally exhaustive algorithm which under the conditions of Theorem 1 is guaranteed to achieve an $\epsilon$-correct clustering of sequences in $X$ with a high probability. The basic idea behind designing this algorithm is to form a series of numerous statistical hypotheses, each of which can be reliably verified or rejected by the mathematical tools developed in the previous sections. More clearly, we consider all possible ways that a given dataset can be partitioned into a number of non-overlapping clusters. Consequently, for each clustering the purity of its clusters will be checked until a sufficiently good candidate is selected. Before proceeding to the algorithm with more details, let us make the following definitions.

**Definition 6.** A clustering or alternatively a partitioning of data samples in $X$ is shown as $Z \in \{1, 2, \ldots\}^n$, where $Z_i$ denotes the cluster index of the $i$th sequence. Hence, the number of clusters would be the maximum index in $Z$. $Z(\kappa, \alpha)$ is the set of all partitionings which cluster $X$ into $\kappa \geq 1$ non-overlapping clusters, each having at least $\alpha n$ members. Finally, $X\mid_{Z=k} \triangleq \{X_i \mid X_i \in X, Z_i = k\}$ for a given partitioning $Z$.

Algorithm 1 finds an $\epsilon$-correct clustering of $n$ data samples in $X$ with probability at least $1 - \zeta$, given the conditions in Theorem 1 are met:

Evidently, Algorithm 1 spans all possible clusters with the number of partitions from 1 to $\lceil \frac{1}{\alpha} \rceil$. As soon as finding a clustering whose every cluster has a sufficiently small maximal mutual dependence measure, the algorithm terminates and outputs the corresponding clustering. If cannot find any clustering with the above-mentioned property, it simply outputs a null clustering.

The parameter $\alpha$ is user-defined. According to our previous assumptions $K$ should not exceed $\left\lceil \frac{1}{\alpha} \right\rceil$. According to Theorem 1 given that this condition holds, the algorithm will terminate before
Algorithm 1: Exhaustive Search

Inputs: $X \in \{0,1\}^{n \times L}$, $L, \delta, \epsilon$, and $\alpha$, such that $L > L^*_c \triangleq \left(\frac{1-\alpha}{\alpha}\right) \frac{(1+\log \frac{1}{\alpha \epsilon})}{2(1-\epsilon)^2}$

Set $L_0 \leftarrow \min \{L^*_c / (1 - L^*_c^{-1}) L\}$
Set $\tau^* \leftarrow \frac{\alpha(1-\epsilon)\delta^2}{1 - \alpha (L_0 - L^*_c)}$
Set $\kappa \leftarrow 1$

while $\kappa < \left\lceil \frac{1}{\alpha} \right\rceil$ do
    for $\forall Z \in \mathcal{Z}(\kappa, \alpha)$ do
        if $\max_k D_{\text{max}}(X|Z=k; L_0) \leq \tau^*$ then
            Set $Z^* \leftarrow Z$, and
            Terminate the program
        end if
    end for
    Set $\kappa \leftarrow \kappa + 1$
end while

Output: $Z^*$, clustering of sequences in $X$.

reaching $\left\lceil \frac{1}{\alpha} \right\rceil$ and as soon as it reaches an $\epsilon$-correct partitioning of $X$ with probability at least $1 - \zeta$. In the following, we use results from Theorem 2 and related lemmas to prove Theorem 1, which is also the mathematical analysis of our proposed algorithm.

**Proof of Theorem 3.** The proposed algorithm will check for all possible cluster numbers $\kappa$ (starting with $\kappa = 1$). Let us denote the number of clusterings that need to be checked before reaching the correct partitioning, i.e. $J$, by $C$. In general, $C$ is a random variable which obviously satisfies the following inequality:

$$C \leq 1^n + 2^n + \ldots + K^n,$$

which simply indicates $C \leq K^{n+1}$. Let us define the following error events during the execution of our proposed algorithm.

$E_1$: Accepting a non-$\epsilon$-pure clustering of $X$, before reaching the true clustering, i.e. when the number of checked clusterings is less than or equal to $C$.

$E_2$: Denying the correct clustering $J$, given that the algorithm has reached it.

Obviously, probability of the algorithm failure, denoted by $P_E$, can be upper-bounded as

$$P_E = \mathbb{P}\{E_1 \cup E_2\} \leq \mathbb{P}\{E_1\} + \mathbb{P}\{E_2\}.$$ 

From Theorem 2 we know that both probabilities of accepting a non-$\epsilon$-pure partitioning, and denying the correct one are bounded by exponentially-decaying terms. By using the union bound over all non-$\epsilon$-pure clusterings in the first $C$ steps of the algorithm, one can see

$$\mathbb{P}\{E_1\} \leq C \mathbb{P}\{E_1^{(1)}\} \leq K^{n+1} 2^{2L^* (1 + \frac{1}{\alpha \epsilon})} \exp \left(\frac{-2\alpha n L \tau^*}{L_0^2 \alpha^2 L^*_c} \right),$$  

(4)
where by \(E_1^{(1)}\) we mean the error event in denial of a single non-\(\epsilon\)-pure clustering during the algorithm. The last inequality in (4) is a direct consequence of the first argument in Theorem 2, considering the fact that for a non-\(\epsilon\)-correct clustering at least one cluster must not be \(\epsilon\)-pure. We have also used the fact that each cluster has at least \(\alpha n\) samples. From (4), it is evident that if

\[
L > \frac{L_0^{5/2} \log K}{\alpha \tau^2} \equiv 4 \log K \left(\frac{L^* \tau^2 / 2 L_c^* / 2}{\epsilon (1 + \log \frac{1}{\alpha \epsilon})}\right)^2,
\]

then \(\lim_{n \to \infty} P\{E_1\} = 0\). The upper bound on \(P\{E_1\}\) for small \(\epsilon\) can be rewritten as

\[
P\{E_1\} \leq K 4^L \exp \left(-\frac{\alpha n \epsilon^2 (1 + \log \frac{1}{\alpha \epsilon})^2}{4 L^* \tau^2 / 2 L_c^*} (L - L_{\text{min}})\right).
\]

A similar argument can be used to obtain an upper-bound on \(P\{E_2\}\). It should be noted that for \(E_2\) to occur at least one of the clusters must have \(D_{\text{max}} > \tau\). Since the number of clusters at that step of the algorithm is \(K\), one can use the union bound over all possible \(K\) clusters each of which has at least \(\alpha n\) members. More precisely, from the second inequality in Theorem 2, it can be shown that

\[
P\{E_2\} \leq K 2^L \exp \left(-\frac{\alpha n \epsilon^2 (1 + \log \frac{1}{\alpha \epsilon})^2}{4 L^* \tau^2 / 2 L_c^*} + L_c^* \log \frac{L}{L_c^*}\right).
\]

Let us find a lower-bound on \(n\), which guarantees the error rate \(P_E\) to be less than \(\zeta > 0\), given that \(L > L_{\text{min}}\). If \(P\{E_1\}\) and \(P\{E_2\}\) are both less than \(\zeta/2\), then \(P_E\) is less than \(\zeta\). Hence, from (6) and (7), we can find minimum number of samples \(n_{\text{min}}\) required to make the probability of error less than \(\zeta\). Clearly, \(n_{\text{min}} \triangleq \max \{n_{\text{min}}^{(1)}, n_{\text{min}}^{(2)}\}\), where \(n_{\text{min}}^{(i)}\) is the minimum number of samples required to achieve \(P\{E_i\} \leq \zeta/2\) for \(i = 1, 2\). From (6) and (7) one can deduce respectively,

\[
n_{\text{min}}^{(1)} \triangleq \frac{4 L^* \tau^2 / 2 L_c^*}{\alpha \epsilon^2 (1 + \log \frac{1}{\alpha \epsilon})^2} \left(\frac{1 + \log \frac{2K}{\zeta} / L}{1 - L_{\text{min}} / L}\right).
\]

and

\[
n_{\text{min}}^{(2)} \triangleq \frac{4 L^* \tau^2 / 2 L_c^*}{\alpha \epsilon^2 (1 + \log \frac{1}{\alpha \epsilon})^2} \left(1 + \log \frac{L}{L_c^*} + \frac{1}{L_c^*} \log \frac{4K^2 L_c^*}{\zeta}\right).
\]

This completes the proof.

The proposed algorithm and Theorem 1 show that as long as the number of informative Bernoulli trials \(L\) is greater than a known threshold, i.e. \(L_{\text{min}} (K, \delta, \epsilon)\), \(\epsilon\)-correctness of clustering is guaranteed almost surely when \(n\) is sufficiently large. In another asymptotic regime, when the total number of Bernoulli trials becomes very large while \(L\) is being fixed, the minimum required dataset size grows logarithmically with \(L\). This phenomenon sounds logical since introduction of more non-informative dimensions in limited \(n\) regimes only adds extra noise and would make the decision-making more challenging.
5 Conclusions

To derive a set of sufficient conditions under which reliable clustering/learning of a BMM is achievable, we have proposed a novel statistic called *maximal mutual dependence measure (m-MDM)* which is computed over a subset of samples and a collection of $L$ dimensions. We have shown that empirical values of m-MDM over a sufficiently pure subset of samples takes small values with high probability. Pureness of a subset of samples reflects the fact that the majority of samples are generated from a particular Bernoulli model of the underlying BMM. On the other hand, large empirical values of m-MDM are reliable indicators of impurity, meaning that samples are more likely to be drawn from different components of the mixture model. This analytic observation has been employed as the main criterion for performing a series of hypothesis tests on all possible ways of partitioning the dataset. Our reliability analysis stipulates certain constraints on the number of samples in the dataset $n$, and also the dimension size $L$, each of which are disclosed in Theorem 1 of the paper. No restrictive assumptions have been made in our model, except those required for the learnability of the problem, such as: sufficient deviation between frequency vectors of different mixture components, and sufficient size of the smallest cluster in the dataset. As a result, our findings encapsulate many classes of BMM inference problems, including those require the independence assumption among frequencies, and also others which take admixture effects into account.

The proposed algorithm in Section 4 is NP-hard with respect to both $n$ and $L$, and thus impractical in real-world situations. However, a goal of this work would be to inspire theoretically plausible heuristics for model-based clustering of BMMs. Looking for more practical and yet theoretically justifiable algorithms in this area can be the focus of our future works. Also, deriving converse conditions for the same problem could be of paramount interest.

A Auxiliary Lemmas and Proofs

In this appendix, we provide the reader with auxiliary Lemmas 3 and 4, and also the proofs of Lemmas 1 and 2, and Theorem 2.

*Proof of Lemma 1.* First, we prove the results in the asymptotic case, i.e. $P\{\lim_{n \to \infty} \mathcal{D}(\mathbf{Y}) \leq 2\tau\} = 0$. Then, we will show that the probability of $|\mathcal{D}(\mathbf{Y}) - \lim_{n \to \infty} \mathcal{D}(\mathbf{Y})| > \tau$ drops exponentially in terms of $n$, which gives us the desired result.

Let $\mathbb{P}_\theta$ to represent the probability distribution of a Bernoulli model with frequency vector $\theta$. Based on the discussion made prior to Lemma 1, one can write

$$\lim_{n \to \infty} \mathcal{D}(\mathbf{Y}) = \mathcal{D}_{KL} \left( \sum_k \phi_k \mathbb{P}_p^{(k)} \right),$$

(9)

where $p^{(k)}$ denotes the frequency vector of the $k$th mixture component, and $\mathbb{E}_k p^{(k)} \triangleq \sum_k \phi_k p^{(k)}$. Here, $\sum_k \phi_k \mathbb{P}_p^{(k)}$ indicates a mixture of Bernoulli models (a BMM), while $\mathbb{P}_{\mathbb{E}_k p^{(k)}}$ denotes a single Bernoulli model whose frequency vector is a weighted average of the $K$ frequency vectors.

It can be clearly seen that when $\phi$ is a delta function, i.e. it is 1 for one $k$ and 0 for the rest, then the two probability distributions $\sum_k \phi_k \mathbb{P}_p^{(k)}$ and $\mathbb{P}_{\mathbb{E}_k p^{(k)}}$ become equal. However, when for all $k$ and
some $\epsilon > 0$ we have $\phi_k \leq 1 - \epsilon$, it can be shown that $\sum_k \phi_k \mathbb{P}_{p^{(k)}}$ cannot be consistently approximated by a single Bernoulli model with parameter $\mathbb{E}_k p^{(k)}$, given that the number of Bernoulli trials $L$ is sufficiently large.

According to the definition of Kullback-Liebler divergence, r.h.s. of (9) can be expanded as follows which helps us to find a proper lower-bound for $\lim_{n \to \infty} D(Y)$:

$$D_{KL} \left( \sum_k \phi_k \mathbb{P}_{p^{(k)}} \| \mathbb{P}_{\mathbb{E}_k p^{(k)}} \right) = \sum_{\forall R} \sum_k \phi_k \mathbb{P}_{p^{(k)}} \{ R \} \log \left( \frac{\sum_u \phi_u \mathbb{P}_{p^{(u)}} \{ R \}}{\mathbb{P}_{\mathbb{E}_k p^{(k)}} \{ R \}} \right) \geq \sum_{\forall R} \sum_k \phi_k \mathbb{P}_{p^{(k)}} \{ R \} \log \left( \frac{\phi_k \mathbb{P}_{p^{(k)}} \{ R \}}{\mathbb{P}_{\mathbb{E}_k p^{(k)}} \{ R \}} \right)$$

$$= \sum_k \phi_k \sum_{\ell=1}^{L} \left( \sum_{r \in \{0,1\}} \mathbb{P}_{p^{(k)}} \{ r \} \log \left( \frac{\mathbb{P}_{p^{(k)}} \{ r \}}{\mathbb{P}_{\mathbb{E}_k p^{(k)}} \{ r \}} \right) \right) - H(\phi)$$

$$= \sum_{\ell=1}^{L} \sum_{k=1}^{K} \phi_k D_{KL} \left( \mathbb{P}_{p^{(k)}} \| \mathbb{P}_{\mathbb{E}_k p^{(k)}} \right) - \mathbb{H}(\phi), \quad (10)$$

where $\mathbb{H}(\phi) \triangleq - \sum_k \phi_k \log \phi_k$ denotes the Shannon entropy of distribution $\phi$, and $\forall R$ stands for $\forall \mathbb{R} \in \{0,1\}^L$. Moreover, it is easy to show that

$$\sum_{k=1}^{K} \phi_k D_{KL} \left( \mathbb{P}_{q_i^{(k)}} \| \mathbb{P}_{\mathbb{E}_k q_i^{(k)}} \right) = H \left( \sum_{k=1}^{K} \phi_k p_{\ell}^{(k)} \right) - \sum_{k=1}^{K} \phi_k H \left( p_{\ell}^{(k)} \right).$$

where $H(p) \triangleq \mathbb{H}(\text{Bern}(p)) = -p \log p - (1-p) \log (1-p)$. Since $H(\cdot)$ is a strictly concave function, and considering the fact that $\mathbb{H}(\phi)$ is bounded from above regardless of $L$, one can conclude that the lower-bound for $\lim_{n \to \infty} D(Y)$ in (10) becomes strictly positive when $i$) $L$ is sufficiently large, and $ii)$ there exist sufficient deviations among $p^{(k)}$s.

In order to simplify the lower-bound, let us assume a random variable $A \in [0,1]$, and define $a = A - \mathbb{E}A$. According to Taylor’s series expansion theorem one can write

$$H(\mathbb{E}A) - \mathbb{E}H(A) = H(\mathbb{E}A) - \mathbb{E} \left\{ H(\mathbb{E}A) + H'(\mathbb{E}A) a + \frac{1}{2} H''(\mathbb{E}A + \xi) a^2 \right\}$$

$$\geq \frac{\mathbb{E}a^2}{2} \inf_{0 \leq p \leq 1} \left| H''(p) \right| = \inf_{0 \leq p \leq 1} \frac{\mathbb{E}a^2}{2p(1-p)} = 2\sigma_A^2, \quad (11)$$

where $\xi$ is a random variable depending on $A$ [11]. Using this inequality, the lower-bound for $\lim_{m \to \infty} D(Y)$ can be written as

$$\lim_{m \to \infty} D(Y) \geq 2 \sum_{\ell=1}^{L} \sum_{k=1}^{K} \phi_k \left( p_{\ell}^{(k)} - \sum_{u=1}^{K} \phi_u p_{u\ell}^{(u)} \right)^2 - \mathbb{H}(\phi). \quad (11)$$

We have already assumed that $\phi_k \leq 1 - \epsilon, \forall k$. Also, for all pairs of rows in $Y$, say $i$ and $j$, there exists a subset of columns $C_{i,j} \subseteq \{1,2,\ldots,L\}$ where

$$\left| p_{\ell}^{(i)} - p_{\ell}^{(j)} \right| \geq \delta, \ell \in C_{i,j} \subseteq \{1,2,\ldots,L\},$$

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and $|C_{i,j}| \geq \mathcal{L}$. Lemma 3 in Appendix A will show that the lower-bound in (11) can be further bounded as

$$
\lim_{n \to \infty} D(Y) \geq 2\mathcal{L} \epsilon \delta^2 (1 - \epsilon) - \mathbb{H}\left(1 - \epsilon, \frac{\epsilon}{K-1}, \ldots, \frac{\epsilon}{K-1}\right) \\
\geq \epsilon \left(2\mathcal{L} (1 - \epsilon) \delta^2 - \left(1 + \log \frac{K}{\epsilon}\right)\right) = 2\tau,
$$

where by choosing $\mathcal{L}$ strictly greater than $\frac{L_c}{K-1} = \frac{1+\log K}{2(1-\epsilon)\delta}$, $\tau$ becomes strictly positive.

However, $D(Y)$ is computed over a finite sample size of $n$, thus it is necessary to show that the estimation error drops sufficiently fast with respect to the size of $Y$. Lemma 4 in Appendix A shows the estimation error in computing $\lim_{n \to \infty} D(Y)$, i.e. $D(Y) - \lim_{n \to \infty} D(Y)$, decays exponentially with respect to $n$. Based on this result, the probability $P\{D(Y) \leq \tau\}$ can be upper-bounded by

$$
P\{D(Y) \leq \tau\} \leq 2L^2 \exp\left(-\frac{n\epsilon^2 (1 - \epsilon)^2 \delta^4}{L^4} \left(L - \frac{L_c}{K-1}\right)^2\right),
$$

which completes the proof.

**Lemma 3.** The lower-bound for $\lim_{n \to \infty} D(Y)$ given in (11), subject to $\phi_k \leq 1 - \epsilon, \forall k$ is as follows:

$$
\lim_{n \to \infty} D(Y) \geq 2\mathcal{L} \epsilon (1 - \epsilon) \delta^2 - \epsilon \left(1 + \log \frac{K}{\epsilon}\right).
$$

**Proof.** Based on the assumption made in Lemma 1 with respect to non-$\epsilon$-purity of $Y$, let us define a subset of $\mathbb{R}^K$ denoted by $\Phi(x)$ for $\epsilon \leq x \leq 1 - 1/K$ as

$$
\Phi(x) \triangleq \left\{\phi \in \mathbb{R}^K \mid \max_k \phi_k = 1 - x, \sum_{k=1}^K \phi_k = 1, \forall \phi_k \geq 0\right\}.
$$

Hence, according to (11) the lower-bound for $\lim_{n \to \infty} D(Y)$ (with $Y$ not being $\epsilon$-pure) can be written as

$$
\lim_{n \to \infty} D(Y) \geq \inf_{\epsilon \leq x \leq 1 - \frac{1}{K}} \inf_{\phi \in \Phi(x)} 2 \sum_{\ell=1}^L \sum_{k=1}^K \phi_k \left(p^{(k)}_{\ell} - \mu_{\ell}(\phi)\right)^2 - \mathbb{H}(\phi),
$$

with $\mu_{\ell}(\phi) \triangleq \sum_{u=1}^K \phi_u p^{(u)}_{\ell}$. In fact, (12) indicates minimization of the lower-bound over all asymptotically large non-$\epsilon$-pure matrices $Y$. In order to further simplify the problem, minimization over $\phi \in \Phi(x)$ can be done for the two terms of r.h.s. of (12), separately. In this regard, it is easy to see that

$$
\sup_{\phi \in \Phi(x)} \mathbb{H}(\phi) = (1 - x) \log \frac{1}{1 - x} + \sum_{k=2}^K \frac{x}{K-1} \log \frac{K-1}{x} \leq x \left(1 + \log \frac{K}{x}\right).
$$
And for the first term in r.h.s of (12), the following lower-bound can be obtained:

\[
\inf_{\phi \in \Phi(x)} 2 \sum_{k=1}^{K} \sum_{\ell=1}^{L} \phi_k \left( p^{(k)}_\ell - \mu_\ell (\phi) \right)^2 \geq \inf_{\phi \in \Phi(x)} 2 \sum_{\ell=1}^{L} \sum_{k=1}^{K} \phi_k \left( p^{(k)}_\ell - \mu_\ell \right)^2
\]

\[
\geq 2 \min_{1 \leq k_0 \leq K} \sum_{\ell=1}^{L} \inf_{\mu_\ell \in \mathbb{R}} \left( 1 - x \right) \left( p^{(k_0)}_\ell - \mu_\ell \right)^2 + \min_k x \left( p^{(k)}_\ell - \mu_\ell \right)^2.
\]

Since for all \( k \) and \( k_0 \), at least \( L \) loci out of \( \ell = 1, 2, \ldots, L \) the inequality \( | p^{(k)}_\ell - p^{(k_0)}_\ell | \geq \delta \) holds, one can write

\[
\inf_{\phi \in \Phi(x)} 2 \sum_{k=1}^{K} \sum_{\ell=1}^{L} \phi_k \left( p^{(k)}_\ell - \mu_\ell (\phi) \right)^2 \geq 2L x (1 - x) \delta^2.
\]

Combining (13) with (14), the following lower-bound can be achieved for \( \lim_{m \to \infty} D(Y) \), which is in terms of the free parameter \( \epsilon \leq x \leq 1 - 1/K \):

\[
\lim_{n \to \infty} D(Y) \geq \inf_{\epsilon \leq x \leq 1 - 1/K} 2L x (1 - x) \delta^2 - x \left( 1 + \log \frac{K}{x} \right).
\]

By taking derivatives from the above lower-bound with respect to \( x \), it can be easily seen that given \( \mathcal{L} > \frac{L}{K-1} \), the minimum occurs for \( x = \epsilon \), which also completes the proof. \( \square \)

**Lemma 4.** The estimation error in computing \( \lim_{n \to \infty} D(Y) \) of Lemma 1 over a finite sample size of \( n \) can be upper-bounded as

\[
P \left\{ \left| D(Y) - \lim_{n \to \infty} D(Y) \right| > \epsilon \right\} \leq 2^{L+1} \exp \left( -\frac{n \epsilon^2}{L^4 2^{L+1}} \right).
\]

**Proof.** For the ease of notation, let us define \( g \left( \hat{p} \right) \triangleq D(Y) \), where \( \hat{p} \) is a \( 2^L \)-dimensional random variable which represents the empirical distribution of the rows of random matrix \( Y \) and \( g : \mathbb{R}^{2^L} \to \mathbb{R}^+ \) to be a corresponding function. In this regard, we employ the indexing form of \( \hat{P}_Y \)'s for \( Y \in \{0,1\}^L \) in order to refer to different dimensions of \( \hat{P} \). According to Definition 3 and due to definition of Kullback-Leibler divergence, it can be readily verified that

\[
D(Y) = \sum_{\forall Y} \hat{P}_Y \log \left( \frac{\hat{P}_Y}{\prod_{\ell=1}^{L} \hat{P}_Y (1 - \hat{p}_\ell)^{1 - Y_\ell}} \right)
\]

\[
= \sum_{\forall Y} \hat{P}_Y \log \hat{P}_Y - \sum_{\forall Y} \hat{P}_Y \sum_{\ell=1}^{L} \left( \log (\hat{p}_\ell) \mathbb{I}(Y_\ell = 1) + \log (1 - \hat{p}_\ell) \mathbb{I}(Y_\ell = 0) \right)
\]

\[
= \sum_{\forall Y} \hat{P}_Y \log \hat{P}_Y - \sum_{\ell=1}^{L} \sum_{\forall Y} \hat{P}_Y \log \left( \sum_{\forall Y' | Y'_\ell = Y_\ell} \hat{P}_Y' \right), \quad (15)
\]
where $I(\cdot)$ denotes the indicator function which returns 1 if its input argument holds and zero otherwise. In derivation of (15) we have used the facts that $\hat{p}_t = \sum_{Y' \mid Y'_t = 1} \hat{P}_{Y'}$ and $1 - \hat{p}_t = \sum_{Y' \mid Y'_t = 0} \hat{P}_{Y'}$.

Noting that $g$ is a continuous function, and also the use of law of large numbers imply that

$$\lim_{n \to \infty} D(Y) = g \left( \lim_{n \to \infty} \hat{P} \right) = g (P^*),$$

where $P^*$ is the true distribution of random Bernoulli vectors generated by the BMM, i.e. $\mathbb{B}$. Note that unlike $\hat{P}$, $P^*$ is a deterministic vector and can be computed given that parameters of $\mathbb{B}$ are known. In this regard, using differential calculus and the mean value theorem, the estimation error can be written and bounded as

$$\left| D(Y) - \lim_{n \to \infty} D(Y) \right| = \left| g \left( \hat{P} \right) - g (P^*) \right| = \left| \int_{\mathcal{P}} \nabla g \cdot d\mathbb{P} \right|$$

$$\leq \sum_{Y \in \{0,1\}^L} \left| \int_{\mathbb{P}^Y \cap \mathcal{P}} \frac{\partial g}{\partial \mathbb{P}^Y} \, d\mathbb{P} \right|$$

$$\leq \sum_{Y \in \{0,1\}^L} \left( \sup_{x \in \mathbb{B}(\mathbb{P}^*, \gamma)} \left| \frac{\partial g}{\partial \mathbb{P}^Y} (x) \right| \right) \left| \mathbb{P}^Y - P^*_Y \right|$$

where $\mathcal{P}$ denotes a path in $\mathbb{R}^{2^L}$ from $P^*$ to $\hat{P}$, and $\mathbb{B}(\mathbb{P}^*, \gamma)$ indicates a ball with radius $\gamma > 0$ around $P^*$ where $\lim_{n \to \infty} \gamma = 0$. Therefore, for sufficiently large $n$, $\gamma$ becomes asymptotically small. According to (15), it is easy to show that partial derivatives of $g$ can be exactly computed through the following formula:

$$\frac{\partial g}{\partial \mathbb{P}^Y} = \log \left( \frac{\hat{P}_Y}{\prod_{t=1}^L \left( \sum_{Y' \mid Y'_t = Y_t} \hat{P}_{Y'} \right)} \right) - (L - 1) \quad \forall Y \in \{0,1\}^L.$$
An important thing which should be noted is that for cases of $P^*_Y = 0$ or $1$, the empirical probabilities $\hat{P}_Y$ always coincide with the true one and their corresponding error terms become exactly zero. Hence such cases are implicitly omitted from all summations.

It is easy to show that the summation over $\forall Y$ in the above inequality reaches its maximum at $P^*_Y = 2^{-L}$ for all $Y$, i.e.

$$\sum_{\forall Y} \left( \log \frac{1}{P^*_Y} + 1 \right) \sqrt{P^*_Y \left( 1 - P^*_Y \right)} \leq L 2^{L/2}.$$ 

What remains to do is to bound the deviations between true distribution $P^*$ and the empirical one $\hat{P}$. Let us define the set of events $A_Y$, $\forall Y \in \{0, 1\}^L$ as

$$A_Y : \left| \frac{\hat{P}_Y - P^*_Y}{\sqrt{P^*_Y \left( 1 - P^*_Y \right)}} \right| > \frac{\varepsilon}{L^{2} 2^{L/2}} \triangleq \delta.$$ 

By using the union and Chernoff bounds, one can have

$$\mathbb{P} \left\{ \left| D(Y) - \lim_{n \to \infty} D(Y) \right| > \varepsilon \right\} \leq \mathbb{P} \left\{ \bigcup_{\forall Y} A_Y \right\} \leq \sum_{\forall Y} \mathbb{P} \left\{ A_Y \right\}$$

$$\leq \sum_{\forall Y} e^{-nD_{KL}(P^*_Y + \delta \sqrt{P^*_Y \left( 1 - P^*_Y \right)} \| P^*_Y)} \mathbb{I} \left( \delta \leq \frac{1 - P^*_Y}{\sqrt{P^*_Y \left( 1 - P^*_Y \right)}} \right)$$

$$+ \sum_{\forall Y} e^{-nD_{KL}(P^*_Y - \delta \sqrt{P^*_Y \left( 1 - P^*_Y \right)} \| P^*_Y)} \mathbb{I} \left( \delta \leq \frac{P^*_Y}{\sqrt{P^*_Y \left( 1 - P^*_Y \right)}} \right),$$

where $\mathbb{I}(\cdot)$ is the indicator function defined before. $D_{KL}(\cdot \| \cdot)$ represents the Kullback-Leibler divergence, where by $D_{KL}(x\|y)$ for $x, y \in [0, 1]$ we simply mean

$$x \log \frac{x}{y} + (1 - x) \log \frac{1 - x}{1 - y}.$$ 

KL divergence measures in the above upper-bounds can be lower-bounded according to Chernoff’s theorem and the probability of observing a deviation greater than $\varepsilon$ in estimating $\lim_{n \to \infty} D(Y)$ can be upper-bounded as

$$\mathbb{P} \left\{ \left| D(Y) - \lim_{n \to \infty} D(Y) \right| > \varepsilon \right\} \leq 2^{L+1} \exp \left( \frac{-n\varepsilon^2}{L^4 2^{L+1}} \right),$$

which completes the proof.\H"{o}lder

Proof of Lemma 2 Proof is highly similar to that of Lemma 1 and 4. The main difference lies in the fact that when $K = 1$, i.e. a single Bernoulli model, one can easily verify

$$\frac{\partial g}{\partial P_{Y}} \Big|_{\hat{P} = P^*} = \log \left( \frac{P_{Y}}{\prod_{\ell=1}^{L} \left( \sum_{\forall Y' \mid Y' \neq Y_{\ell}} P_{Y'} \right)} \right) - (L - 1) = 1 - L,$$

$\forall Y \in \{0, 1\}^L$.\end{proof}
and therefore
\[ \left| \frac{\partial g}{\partial \hat{P}_Y} \right| \bigg|_{\hat{P}=P} \leq L. \]

Then, following the same steps as shown in proof of Lemma 4 will complete the proof.

**Proof of Theorem 2.** The first inequality states that the probability of all \( \binom{L}{L_0} \) sub-matrices of \( Q \) with \( L_0 \) columns having \( D < \tau \) is strictly bounded from above. In order to show that, note that frequency matrix \( p \in [0, 1]^{K \times L} \) is assumed to be \((L, \delta)-separable\), which means for every pair of rows in \( p \) there are at least \( L \leq L \) Bernoulli trials where their frequencies differ in a value greater than or equal to \( \delta > 0 \) between the two clusters.

It can be easily seen that by examining all \( \binom{L}{L_0} \) subsets of loci with size \( L_0 \), there are at least \( h \triangleq \left\lfloor \frac{L}{L_0} \right\rfloor \) non-overlapping sub-matrices of \( Q \), denoted by \( S_1, \ldots, S_h \), which are \((L_0/(K - 1), \delta)-separable\). Since \( S_1, \ldots, S_h \) do not overlap with each other, they are statistically independent which implies

\[
\mathbb{P}\{D_{\max}(Y, L_0) \leq \tau\} \leq \prod_{i=1}^{h} \mathbb{P}\{D(S_i) \leq \tau\} = (\mathbb{P}\{D(S_1) \leq \tau\})^h.
\]

Using the upper-bound for \( \mathbb{P}\{D(S_1) \leq \tau\} \) which is derived in Lemma 4 and approximating \( h \) with \( L/L_0 \) provide us with the claimed inequality.

For the case of second inequality, one can simply employ union bound as

\[
\mathbb{P}\{D_{\max}(Y; L_0) > \tau\} \leq \mathbb{P}\left\{ \max_{S \in \text{col}(Y; L_0)} D(S) > \tau \right\} \leq \sum_{S \in \text{col}(Y; L_0)} \mathbb{P}\{D(S) > \tau\},
\]

where \( \text{col}(Y; L_0) \) represents the set of all \( \binom{L}{L_0} \) sub-matrices of \( Y \) with \( L_0 \) columns. Again, substitution of \( \mathbb{P}\{D(S) > \tau\} \) with the upper-bound derived in Lemma 4 gives us the claimed inequality and will complete the proof.

**References**

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\[\text{In practice, since informative loci are dispersed randomly among all Bernoulli trials, \((L_0, \delta)\)-separability holds almost surely for sufficiently large } L_0. \text{ However, we have considered the improbable worst case where only } (L_0/(K - 1), \delta)\)-separability is guaranteed.\]
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