RECONSTRUCTING PEDIGREES: A STOCHASTIC PERSPECTIVE

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Abstract. A pedigree is a directed graph that describes how individuals are related through ancestry in a sexually-reproducing population. In this paper we explore the question of whether one can reconstruct a pedigree by just observing sequence data for present day individuals. This is motivated by the increasing availability of genomic sequences, but in this paper we take a more theoretical approach and consider what models of sequence evolution might allow pedigree reconstruction (given sufficiently long sequences). Our results complement recent work that showed that pedigree reconstruction may be fundamentally impossible if one uses just the degrees of relatedness between different extant individuals. We find that for certain stochastic processes, pedigrees can be recovered up to isomorphism from sufficiently long sequences.

1. Introduction

Since earliest civilisation people have been concerned with recording, deciphering and resolving their ancestry. The concept of a ‘family tree’ is widely familiar (even though the ancestry of an individual cannot remain a tree for too many generations into the past) and there are many methods for deciphering ancestry back several generations. Mostly these are somewhat ad-hoc, based on comparing and combining overlapping ancestries, oral and written records.

However in recent decades the concept of deeper ancestry has become topical in molecular evolution. Firstly, the ‘Out-of-Africa’ hypothesis [1], now widely accepted, suggests that all extant humans are descendants of a relatively small population that migrated (possibly multiple times) out of Africa around 150,000-200,000 years ago. Secondly, recent theoretical work [7] suggests that most of the human population is likely to have common ancestors much more recently (thousands rather than hundreds of thousands of years ago). Thirdly, since the sequencing of the complete human genome in 2001, [3, 11] and subsequent

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improvements in the economics and speed of sequencing technology, it is quite possible that complete (or near-complete) genomic sequences for all individuals in a population could be available in the near future.

These factors immediately suggest the question: what would a very large amount of genomic data tell us about the ancestry of a population? Clearly one can easily decide who are closely related (siblings, cousins etc), but how far back in time might one be able to reconstruct an accurate ancestry? To date, little is known about what is needed in order to formally reconstruct a pedigree (a graph that describes ancestry – defined formally below) though some initial results were presented in [8]. This is in marked contrast to another field in molecular evolution, namely phylogenetics, where there is a well-developed theory for reconstructing evolutionary (‘phylogenetic’) trees on species from the genetic sequences of present-day species [4]. In that setting genetic data is often highly informative for reconstructing detailed relationships between species deep into the past (tens or hundreds of millions of years). They can also be informative at short time frames when studying rapidly evolving organisms (such as HIV).

However in phylogenetics the underlying graph is a tree, while in a pedigree it is a more ‘tangled’ type of directed graph. Moreover, the number of vertices in a tree is linearly related to the number of leaves (which represent the extant species on which we have information) while for a pedigree the number of vertices (individuals) can keep growing as we go further back in time.

In this paper we continue the analysis started in [8] and attempt to determine models under which pedigrees might be reconstructed from sufficient data. We should point out that there is a well-developed statistical theory for pedigrees [10], but this deals with different sorts of questions than pedigree reconstruction, such as estimating an ancestral state in a known pedigree.

In [8] and [9], pedigrees were considered mainly from a combinatorial perspective. A question considered in both these papers was how best to construct pedigrees from certain combinatorial information about them, such as sets of distances between individuals, pedigrees on sub-populations, and so on. Several examples and counterexamples to combinatorial identifiability questions were presented. It seemed that constructing pedigrees would be a difficult task, if at all possible, and some of our intuition derived from phylogenetic trees would not carry over to pedigrees.

A purpose of this paper is to consider pedigrees from a more stochastic perspective. We consider several stochastic models of evolution on a pedigree, that is, mechanisms by which individuals may inherit
sequence information from their parents. We consider the fundamental theoretical question: is the sequence information available in living individuals in a population sufficient to construct the pedigree of the population, or might there instead be portions of a pedigree, that will always remain ghosts, unable to be clearly resolved regardless of how much sequence data one has on extant individuals? More formally, we are interested in whether non-isomorphic pedigrees could produce the same joint distribution of sequence information for living individuals. We begin with some combinatorial preliminaries and enumerate the number of distinct pedigrees to strengthen an earlier lower bound on the number of segregating sites that was derived in [8].

2. Definitions and preliminaries

Mostly we follow the notation of [9]. Unless stated otherwise we will assume all (directed or undirected) graphs are finite, simple and without loops. A general pedigree is a directed acyclic graph $P = (V, A)$ in which $V$ can be written as the disjoint union of two subsets $M$ and $F$ (‘Male’ and ‘Female’), and where each vertex either has no-incoming arc or two incoming arcs, with one from a vertex in $M$ and the other from a vertex in $F$. The vertices with no in-coming arcs are called the founder vertices.

In representing ancestry an arc $(u, v)$ of $P$ denotes that $v$ is a child (offspring) of $u$ (equivalently, $u$ is a parent of $v$), and the conditions defining a pedigree simply state that each individual (not in the founding population) has a male and female parent, and that there is an underlying temporal ordering (acyclicity).

In Figure 1, a general pedigree is shown on the left.

Given a directed graph $G = (V, A)$ let $M(G) = (V, E)$ be the graph on $V$ whose edge set consists of all pairs $\{u, v\}$ for which there exists $w \in V$ with $(u, w) \in A$ and $(v, w) \in A$. In the case where $G$ is a ‘food web’, $M(G)$ is known as the ‘competition graph’ (see [6]). However in our setting, if $G$ is a pedigree, then $M(G)$ is the ‘mate graph’ of $G$, where a pair of individuals form an edge if they have at least one child.

Lemma 1. A directed graph $G = (V, A)$ is a pedigree if and only if (i) $G$ is acyclic, (ii) $M(G)$ is bipartite, and (iii) no vertex of $G$ has just one incoming arc. In particular it can be determined in polynomial time (in $|V|$) whether or not a directed graph is a pedigree.

Proof. Conditions (i)–(iii) clearly hold if $G$ is a pedigree. Conversely, if $M(G)$ is bipartite $V$ can be properly 2-coloured, with colour set $\{M, F\}$, and so we can write $V$ as the disjoint union of two sets $M, F$ so that each vertex with at least two incoming edges has exactly two
incoming edges – one from a vertex in $M$ and one from a vertex in $F$. Condition (iii) excludes the possibility of just one incoming edge, and so $G$ is a pedigree. For the second claim, observe that the three conditions (i)–(iii) can all be established in polynomial time. □

The set of vertices that have no out-going arcs is denoted $X_0$, and for a particular distinguished subset $X$ of $X_0$ (called the extant individuals) we refer to $(P, A)$ as a pedigree on $X$. We assume that the vertices in $X$ are labelled, and other vertices are unlabelled. Two pedigrees on $X$ are isomorphic if there is a diagraph isomorphism between them that fixes each element of $X$.

We note in passing that in [8] it was sometimes assumed that the decomposition $(M, F)$ of $V$ was known, as this is not necessarily uniquely determined just by $P$; this in turn also allows a more restrictive definition of isomorphism (called ‘gender-isomorphism’) in which the diagraph isomorphism is required to map $M$ (resp. $F$) vertices to $M$ (resp. $F$) vertices. However we do not require or invoke this additional structure in the current paper.

A simple pedigree is a pedigree in which the vertex set of the pedigree is a disjoint union of $X_i; 0 \leq i \leq d$, and every arc $(u, v)$ has its tail $u$
in $X_i$ and its head $v$ in $X_{i-1}$, for some $i > 0$. In this case, $X_0$ is the set of extant vertices, and $X_d$ is the set of founders, and $d$ is the depth of the pedigree. In \cite{8} and \cite{9}, the term ‘discrete generation pedigree’ was used instead of the term ‘simple pedigree’. In simple pedigrees with a constant population size, all $X_i$ have the same cardinality. In Figure \ref{fig:pedigree}, a simple pedigree with a constant population size is shown on the right.

The amount of information required to accurately reconstruct a pedigree on a set of size $n$, and up to depth $d$, is clearly bounded below by some increasing function of the number of distinct (mutually non-isomorphic) simple pedigrees with a constant population size $n$ and of depth $d$. Let this number be $f(n, d)$. We first describe a lower bound on $f(n, d)$ providing a slightly stronger bound than \cite{8}.

Let $X_0 = \{x_i; 1 \leq i \leq n\}$ and $X_1 = \{y_i; 1 \leq i \leq n\}$. Consider a tree $T$ defined on $X_1$. We construct a pedigree on $X_0 \cup X_1$ with the set of extant vertices $X_0$ as follows: we first take an arbitrary onto map $g$ from $X_0$ to the edge set $E(T)$ of $T$, and for every $x_k \in X_0$, if $g(x_k) = \{y_i, y_j\}$, then in the pedigree, $x_k$ is a child of $y_i$ and $y_j$. We count the number of pedigrees that can be constructed in this manner by considering all possible mutually non-isomorphic trees $T$, and all possible onto maps from $X_0$ to $E(T)$. For a fixed tree $T$, there are exactly $\binom{n}{2}(n-1)!$ onto maps from $X_0$ to $E(T)$. Each map does not give us a distinct pedigree; in fact, each pedigree constructed this way is repeated $|\text{aut} T|$ times, where $\text{aut} T$ is the automorphism group of $T$. Thus we have

$$f(n, 1) \geq \sum_T \frac{\binom{n}{2}(n-1)!}{|\text{aut} T|},$$

where the summation is over all mutually non-isomorphic trees on $X_1$. Since $n!/|\text{aut} T|$ is the number of labelled trees isomorphic to a given tree $T$, summing over all mutually non-isomorphic trees gives us

$$f(n, 1) \geq \frac{(n-1)n^{n-2}}{2},$$

where $n^{n-2}$ is the number of labelled trees on $X_1$, by Cayley’s classic formula \cite{2}.

Observe that each vertex in $X_1$ is distinguished in the pedigree, in the sense that no two vertices in $X_1$ have the same set of children. This fact is useful to construct distinct pedigrees of arbitrary depth by repeating the same construction for arcs between $X_1$ and $X_2$, $X_2$ and $X_3$, . . . . Therefore,
\[ f(n, d) \geq \frac{(n - 1)d n^{d(n-2)}}{2^d} \]

Observe also that, since trees are bipartite, the directed graph constructed is indeed a pedigree by Lemma 1.

The above estimate gives an information theoretic lower bound of \((d/2) \log n + o(\log n)\) on the number of segregating sites needed for reconstructing a pedigree from DNA sequence data. This follows by the same argument as in [8] and is a slight improvement on the bound \((d/3) \log n + o(\log n)\) established in that paper.

3. Pedigree reconstruction

In this section, we examine the question of constructing a pedigree from the information obtained from the extant individuals. In biological applications, this information is typically provided by (DNA) sequence data. It is assumed that the information has been passed on to each individual by its parents; and, over generations, the information undergoes a stochastic change that models the evolutionary process. Is the information available at all extant individuals sufficient to uniquely construct the pedigree of the population? To be precise, are there examples of stochastic processes for which we cannot construct the pedigree, and are there examples of stochastic processes for which we can construct the pedigree?

3.1. A negative result. We begin with a simple Markov process under which the information at the extant vertices (in the form of binary sequences of arbitrary length) is not sufficient to uniquely determine the pedigree.

Suppose \( \{u_i; 1 \leq i \leq p\} \) is the vertex set of a pedigree \( \mathcal{P} \). Suppose that associated with each vertex \( u_i \) in the pedigree \( \mathcal{P} \), there is a random variable \( U_i \) that takes values from a finite state space \( S \). Let

\[ \mathbb{P}(U_i = a_i | U_j = a_j; 1 \leq j \leq p, j \neq i) \]

denote the probability that \( U_i \) takes the value \( a_i \) conditional on the states of random variables at all other vertices. We assume that

\[ \mathbb{P}(U_i = a_i | U_j = a_j; 1 \leq j \leq p, j \neq i) = \mathbb{P}(U_i = a_i | U_j = a_j, U_k = a_k), \]

where \( u_j \) and \( u_k \) are the parents of \( u_i \). Is it possible to construct the pedigree up to isomorphism given the joint distribution \( \mathbb{P}(U_1 = a_1, U_2 = a_2, \ldots, U_n = a_n) \), where we use the indices 1 to \( n \) for extant vertices?

Consider a symmetric two-state model given by the transition matrix
where the columns are indexed by the joint states of the parents of a vertex, and the rows are indexed by the state of the vertex. For example, the entry in the first column and second row says that the probability that a child is in state 1 conditional on both parents being in state 0 is $1 - \alpha$.

In the following, we construct non-isomorphic pedigrees $\mathcal{P}$ and $\mathcal{Q}$, each on two extant vertices $u_1$ and $u_2$, such that the joint distribution $\mathbb{P}(U_1 = a_1, U_2 = a_2)$, where $a_i \in \{0, 1\}$, is identical for $\mathcal{P}$ and $\mathcal{Q}$.

(1) Construct two disjoint binary pedigrees $\mathcal{B}_i; i \in \{1, 2\}$, respectively, on extant vertices $u_1$ and $u_2$. The depth of each binary pedigree is $t \geq 2$. Let $S_i; i \in \{1, 2\}$ be the corresponding sets of their founders.

(2) Construct a single intermediate pedigree $\mathcal{P}'$ from $\mathcal{B}_i; i \in \{1, 2\}$ by identifying each vertex in $S_1$ with a unique vertex in $S_2$. Construct pedigree $\mathcal{P}$ by adding vertices $v$ and $w$ as parents of all founder vertices in the pedigree $\mathcal{P}'$.

(3) Construct pedigree $\mathcal{Q}$ as in the above step so that $\mathcal{P}$ and $\mathcal{Q}$ are non-isomorphic. This is possible when $t \geq 2$.

Figure 2 shows examples of $\mathcal{P}$ and $\mathcal{Q}$ for $t = 2$.

**Proposition 1.** The pedigrees $\mathcal{P}$ and $\mathcal{Q}$ have the same joint distribution $\mathbb{P}(U_1 = a_1, U_2 = a_2)$, where $a_i \in \{0, 1\}$, under the symmetric model described above. Thus the two pedigrees cannot be distinguished from each other from binary sequences (of i.i.d. samples) of any finite (or infinite) length.

**Proof.** First consider a binary pedigree, say $\mathcal{B}_1$. Let $k$ of the vertices in $S_1$ be in state 0. Let $f(k, t)$ denote the probability that the vertex $u_1$ is in state 0. Suppose $k_1$ of the 0 states occur among the founders on the left tree, and $k_2$ occur on the right tree, where the left tree and the right tree are the pedigrees of the two parents of $u_1$. Therefore, $k_1 + k_2 = k$. A recurrence for $f(k, t)$ is then written in terms of $f_1 = f(k_1, t - 1)$ and $f_2 = f(k_2, t - 1)$.

$$f(k, t) = \alpha f_1 f_2 + 0.5(1 - f_1) f_2 + 0.5 f_1 (1 - f_2) + (1 - \alpha)(1 - f_1)(1 - f_2),$$

where the four terms correspond to the four possible joint states of the parents of $u_1$. The proposition holds for $t \geq 2$. Figure 2 shows examples of $\mathcal{P}$ and $\mathcal{Q}$ for $t = 2$. 
Figure 2. Non-isomorphic pedigrees that produce indistinguishable sequences under the symmetric stochastic model.

It can be verified by induction that the following expression for \( f(k, t) \) solves the recurrence.

\[
f(k, t) = \frac{k}{2^t} (2\alpha - 1)^t + 1 - \frac{(2\alpha - 1)^t}{2}.
\]

Here the independence of \( f(k, t) \) on exactly where the zero states occur among the founders is what is useful in the following.

Now consider the intermediate pedigree \( P' \) and consider the event \( E_k \) that exactly \( k \) of its founders are in state 0 (so \( k \in \{0, 1, 2, 3, 4\} \)). The conditional probability \( \mathbb{P}(U_1 = a_1, U_2 = a_2 | E_k) \) is given by

\[
\mathbb{P}(U_1 = a_1, U_2 = a_2 | E_k) = \mathbb{P}(U_1 = a_1 | E_k) \mathbb{P}(U_2 = a_2 | E_k),
\]

where each factors is either \( f(k, t) \) or \( 1 - f(k, t) \) depending on whether \( a_i \) are 0 or 1, respectively. This is also true in \( Q' \).

The vertices \( v \) and \( w \) are added to both intermediate pedigrees as parents of vertices in \( S_1 \) and \( S_2 \) so as to guarantee that all possible joint states on \( S_i \) that have \( k \) zeros are equally likely. This implies that for any given joint distribution on \( v \) and \( w \), we have the same joint distribution on \( u_1 \) and \( u_2 \) in \( P \) and \( Q \).

We now show that exponentially many mutually non-isomorphic pedigrees can be obtained by this construction.
Proposition 2. \textit{The number of mutually non-isomorphic pedigrees that can be obtained by the above construction grows super-exponentially with } $t$.

\textit{Proof.} Consider two disjoint binary pedigrees $B_i$ of depth $t \geq 2$, on extant vertices $u_i$, and founder sets $S_i$, where $i \in \{1, 2\}$. Let $|S_i| = 2^t = m$. There are $m!$ ways of identifying vertices in $S_2$ with vertices in $S_1$, but not all of them result in mutually non-isomorphic pedigrees. Consider a pedigree $P'$ obtained by identifying vertices in $S_2$ with vertices in $S_1$. The automorphism group of $P'$ is a subgroup of the automorphism group of $B_1$. But $|\text{aut } B_1| = 2^{m-1}$, therefore, $|\text{aut } P'| \leq 2^{m-1}$. Therefore, the number of mutually non-isomorphic pedigrees obtained by identifying vertices of $S_2$ with vertices in $S_1$ is at least

$$\frac{m!}{2^{m-1}},$$

which implies the claim. \hfill \Box

3.2. \textbf{Positive results.} We first describe a simple deterministic process, and a related stochastic variation, under which the information available at the extant individuals is sufficient to construct the pedigree. We then describe a Markov model that comes closer to the mutation-recombination setting of genetic ancestry, for which pedigree reconstruction is also possible. This last model should be viewed as a proof-of-concept, rather than as realistic processes that capture all aspects of evolutionary processes.

\textbf{Example 1} (Deterministic process). Suppose each founder in the population has a distinct label. Consider an individual whose parents are labelled $Y$ and $Z$. Suppose that each individual inherits the labels of its parents, and also has its own unique character that is not seen before in any other individual. In this way we assign the individual a label $\{\{Y, Z\}, X\}$, where $X$ is a new symbol or a trait that no other individuals in the population, except for descendents of the individual under consideration, who inherit $X$ in the manner described.

From the labels of the extant individuals, the pedigree is uniquely constructed in a straightforward manner. First we construct the pedigree of each extant individual. Each individual’s label uniquely determines the labels of its parents and the new character that has arisen in the population for the first time. We recursively construct a binary tree of parents, grand parents, ... beginning with an extant individual. After constructing the binary tree, we identify vertices that have the same labels. Such vertices are ancestors to whom there are multiple paths from the extant individual.
The next step is to construct a (graph theoretic) union of pedigrees of all extant individuals. In constructing the graph theoretic union, vertices in different pedigrees that have the same labels are identified, and multiple arcs between two vertices are suppressed to leave a single arc between them. This completes the construction.

**Example 2** (Semi-deterministic process on the integers). Now we modify Example 1 so as to introduce some randomness, and also to work over a fixed state space (the integers). Let $N$ be a large positive integer (sufficiently large relative to the number of vertices in the pedigree, in a sense that will be made more precise shortly). To each individual $i$ in the pedigree we first associate an independent random variable $Y_i$ that takes a value selected uniformly at random from $\{1, \ldots, N\}$. We then assign a random state $X_i$ to each vertex $i$ of the pedigree as follows. If $i$ is a founder, then set $X_i = Y_i$. Otherwise, if $i$ has parents $j$ and $k$ then set

$$X_i = 2^{X_j+N} + 2^{X_k+N} + Y_i.$$ 

Observe that this process is Markovian (the state at a vertex depends just on the states at the parents, and not on earlier ancestors). Moreover, if the random variables $Y_i$ take distinct values, then the pedigree can be uniquely constructed since $2^{a+N} + 2^{b+N} + m$ can be uniquely ‘decoded’ as $\{\{a, b\}, m\}$. If there are $n$ vertices in the pedigree (and $N \geq n$) the probability that each random variable takes a distinct value is

$$\frac{N(N-1)\ldots(N-n+1)}{N^n},$$

which approaches 1 as $N$ tends to infinity.

Therefore, under this process, a pedigree can be uniquely reconstructed by observing the random variables at the extant vertices, with a probability approaching 1 as $N$ tends to infinity.

Although the above examples seems to be far removed from the reality of biological evolution, the concept underlying the examples is almost un-recognisably hidden in the following setting where the main consideration is to construct a process that models sequence evolution.

### 4. A STOCHASTIC PROCESS ON SEQUENCES THAT ALLOWS RECONSTRUCTION

The process of inheriting genetic material from parents may be conceptualised as follows. Suppose the parents $Y$ and $Z$ of an individual $X$ have sequences $\{y_i; i = 1, 2, \ldots\}$ and $\{z_i; i = 1, 2, \ldots\}$, respectively.
Here the sequences are assumed to be sequences of characters drawn from \([N] = \{1, 2, \ldots, N\}\). We assume that the sequence \(\{x_i\}\) of \(X\) is constructed by copying segments of sequences \(\{y_i\}\) and \(\{z_i\}\) so that roughly half the genetic material is inherited from one parent, and roughly half from the other parent. In addition to the directly copied bits and pieces from its parents’ genetic sequences, \(X\) also has in its sequence occurrences of segments that are not (recognised as) copies of segments of \(\{y_i\}\) and \(\{z_i\}\). We suppose that the \(X\)-specific fragments are constructed from characters drawn from a set \(U_X \subset [N]; |U_X| = m\), where \(U_X\) is chosen uniformly at random from the family of all subsets of \([N]\) of cardinality \(m\). The process of construction of the sequence \(\{x_i; i = 1, 2, \ldots\}\) is then modelled as in a hidden Markov model. The copying process copies character from \(\{y_i\}\), and at some step, determined by chance, begins copying characters from \(\{z_i\}\), or begins a random generation of a sequence of characters chosen from \(U_X\). The process of copying from and switching between \(\{y_i\}\), \(\{z_i\}\) and \(U_X\) continues.

But the segments copied from \(\{y_i\}\) and \(\{z_i\}\) are in turn partly inherited from the parents of \(Y\) and \(Z\), respectively, and partly from the \(Y\)-specific and \(Z\)-specific segments, that is, segments of characters drawn from \(U_Y\) and \(U_Z\), respectively.

We model the above description by first defining a one to one correspondence between pedigrees and a subclass of finite automata that emit (to use the HMM terminology) character sequences at the extant individuals. We then demonstrate how a sufficiently long emitted sequence determines first the automaton and then the pedigree with high probability.

Without a loss of generality, we consider pedigrees with a single extant vertex, since after constructing all sub-pedigrees having a single extant vertex, we can construct their graph theoretic union, as in Example 1. This is discussed further in Remark 1.

4.1. The automaton (directed graph) \(G\), and the mechanism of sequence emission. Let \(Q\) be a pedigree with vertex set \(V; |V| = n\), with a single extant vertex \(x\). The automaton associated with \(Q\) is denoted by a directed graph \(G\) on the vertex set \(V\). For convenience, we have used the same vertex set \(V\); so to avoid ambiguity, we denote an arc from \(y\) to \(z\) in \(Q\) by \(yz\), and an arc from \(y\) to \(z\) in \(G\) by \((y, z)\).

The automaton \(G\), its transition probabilities, and the mechanism by which it emits characters in the sequence of the extant vertex are defined so that the following conditions are satisfied.
(1) Let $[\delta_1, \delta_2] \subseteq [0, 1]$ and $[\Delta_1, \Delta_2] \subseteq [0, 1]$ be two intervals such that $\delta_i$ are much smaller than $\Delta_j$ for $i, j \in \{1, 2\}$.

(2) For each internal vertex $y$, (that is, a vertex that is neither a founder vertex nor the extant vertex), there are two arcs $(y, u)$ and $(y, v)$ to its parents $u$ and $v$, respectively, an arc $(y, x)$ to the extant vertex $x$, and a self loop. We assume that the transition probabilities satisfy

$$ p(y, u), p(y, v) \in [\Delta_1, \Delta_2] $$

and

$$ p(y, x), p(y, y) \in [\delta_1, \delta_2]. $$

(3) For the extant vertex $x$, there are outgoing arcs $(x, y)$ and $(x, z)$ to its parents, $y$ and $z$, respectively, and a self-loop, with the corresponding transition probabilities given by

$$ p(x, y), p(x, z) \in [\Delta_1, \Delta_2] $$

and

$$ p(x, x) + p(x, y) + p(x, z) = 1. $$

(4) From a founder vertex $z$, there is one arc $(z, x)$ to the extant vertex $x$, and a self-loop. The transition probabilities satisfy

$$ \delta_1 \leq p(z, x) \leq \delta_2 $$

and

$$ p(z, x) + p(z, z) = 1. $$

(5) Each vertex $y$ of the automaton corresponds to a subset $U_y$ of $[N]$, such that $|U_y| = m > 1$, and $U_y$ is chosen randomly from a uniform distribution on the family of subsets of $[N]$ of cardinality $m$. The character sequence for $x$ is emitted by the automaton as follows: the automaton defines a Markov chain with transition probabilities defined above; when the chain is in state $y$, (that is, at vertex $y$ of the automaton), a character from $U_y$ is emitted from a uniform distribution on $U_y$; $y \in V$.

The assumption that $\delta_i$ are much smaller than $\Delta_j$ for $i, j \in \{1, 2\}$, and the conditions listed above imply that an individual derives most of its genetic material from its parents, who in turn receive most of their genetic material from their parents.

Figure 3 shows a pedigree $Q$ on 6 vertices and an automaton $G$ that corresponds to the pedigree $Q$. The transition probabilities in the figure are denoted by $\Delta_{ij}$ or $\delta_{ij}$ instead of $p(i, j)$ so as to indicate their relative magnitudes.
We are interested in the following question: does a sufficiently long sequence \( \{x_i; i = 1, \ldots \} \) emitted by the automaton determine the pedigree unambiguously with high probability? Since the correspondence between the subclass of automata and pedigrees with a single extant vertex is one-to-one, the question is equivalent to asking if the automaton can be constructed unambiguously. The main result of this section is the affirmative answer to this question, formulated in the following theorem. Note that although it deals with only a single extant vertex, we describe in Remark 1 how it extends to the general case of a pedigree over a finite set \( X \).

**Theorem 1.** Let \( Q \) be a pedigree having a single extant vertex. Let \( Q \) be associated with an automaton \( G \) that satisfies the conditions listed above. Let \( S_k = \{x_i; i = 1, 2, \ldots, k\} \) be a sequence of characters from the set \([N] = \{1, 2, \ldots, N\}\), emitted by the automaton (as in the fifth condition above). Then for sufficiently large \( k \) and \( N \), the automaton \( G \) and the pedigree \( Q \) can be correctly reconstructed (with high probability) from the sequence \( S_k \).

The theorem follows from the several lemmas proved next.
Lemma 2. Given an automaton $G$ with its transition probabilities, the pedigree $Q$ can be uniquely constructed.

Proof. This follows from the relative magnitudes of the probabilities of transition. For distinct vertices $u$ and $v$ in $G$, the transition probability from $u$ to $v$ is high, (that is, in the interval $[\Delta_1, \Delta_2]$), if and only if $v$ is a parent of $u$ in the pedigree $Q$. For a vertex $u$, the probability of transition from $u$ to itself is high if and only if $u$ is a founder vertex. A vertex $u$ is the extant vertex of $Q$ if and only if there is no other vertex $v$ in $G$ such that the probability of transition from $v$ to $u$ is high. □

Next we must construct the automaton $G$ from the sequence $S_k$. The idea of inference of the automaton $G$ from the sequence $S_k$ is based on the following observation. Suppose $i, j \in [N]$ are such that there is only one $U_y$ that contains $i$, and only one $U_z$ that contains $j$. Then the observed transition probability $p(i|j)$ in the sequence $S_k$ is in the range $[\Delta_1/m, \Delta_2/m]$ if $y$ is a parent of $z$; and is in the range $[\delta_1/m, \delta_2/m]$ if $i \in U_z$ and $j \in U_y$, or if $\{i, j\} \subseteq U_y$, where $y$ is an internal vertex. Similarly, one can argue about the magnitude of the observed frequency of $i$ followed by $j$ in $S_k$ for founder vertices, and for the extant vertex. What matters is whether the estimated probability is high (of the order of $\Delta_i/m; i = 1, 2$) or low (of the order of $\delta_i/m; i = 1, 2$). The transition probabilities $p(i|j)$ can be estimated as accurately as desired by choosing sufficiently large $k$. It is crucial for the above argument that each $U_y$ contains some state $i$ that is unique to $U_y$, that is, $i$ does not belong to a $U_z$ for $z$ other than $y$. This is the case with high probability for large $N$, as made precise in the following lemma.

Lemma 3. Suppose that the sets $U_y$ are randomly chosen from a uniform distribution on the family of subsets of $[N]$ of cardinality $m$. Let $E$ be the event that each $U_y$ contains at least one $i$ that is not in any other $U_z$. The probability of this event $E$ approaches 1 as $N$ tends to infinity.

Proof. Let $E_i$ be the event that $U_i$ is not a subset of $\cup_{j \neq i} U_j$. Then, $E = \cap_{i=1}^n E_i$, and by Boole’s inequality $\mathbb{E}$, and symmetry,

$$\mathbb{P}(E) \geq 1 - \sum_{i=1}^n \mathbb{P}(E^c_i) = 1 - n\mathbb{P}(E^c_1),$$

where the superscript $c$ denotes complement. Now $E_1^c$ is the event that $U_1$ is a subset of $U_2 \cup E_3 \cup \ldots \cup U_n$, and clearly the probability of this (complementary) event is maximised if $U_2, \ldots, U_n$ are disjoint. In this case $|U_2 \cup \ldots \cup U_n| = (n - 1)m$, and so $\mathbb{P}(E_1^c)$ is bounded above by the proportion of subsets of $[N]$ of size $m$ that are subsets of a set of size $m$. 


(n - 1)m, i.e. \( \mathbb{P}(E^c_i) \leq \frac{(n-1)m}{(m)} \). This, along with the above inequality, implies \( \mathbb{P}(E) \rightarrow 1 \) as \( N \rightarrow \infty \). \( \square \)

Let \( U_i \subseteq [N]; i = 1, 2, \ldots n \) be the unknown character sets corresponding to the vertices 1, 2, \ldots, n of the automaton. Let \( \bar{U}_i \) denote the subset of \( U_i \) consisting of those elements that are unique to \( U_i \), that is,

\[ \bar{U}_i = U_i \cap (\cup_{j \neq i} U_j)^c. \]

By a recursive procedure, we construct \( \bar{U}_i \), and the pedigree \( Q \) on the vertex set \([n] = \{1, 2, \ldots, n\}\).

Without a loss of generality, assume that the extant vertex is labelled 1, and the founder vertices are labelled from \( f \) to \( n \).

We first construct a directed graph \( H \) from the observed sequence \( x_i; i = 1, 2, \ldots \). The vertex set \( V(H) \) of \( H \) is the set of states that appear in the emitted sequence \( x_i; i = 1, 2, \ldots \). The set of arcs of \( H \) is \( E(H) \), and an arc \( (u, v) \) is in \( E(H) \) if a transition from \( u \) to \( v \) is observed in \( x_i; i = 1, 2, \ldots \), that is, if there is some \( i \) for which \( x_i = u \) and \( x_{i+1} = v \). Each arc \( (u, v) \) of \( H \) is labelled high or low depending on whether the inferred probability \( p(v|u) \) of transition from \( u \) to \( v \) is of the order of \( \Delta/m \) or \( \delta/m \), respectively, where \( \Delta_1 \leq \Delta \leq \Delta_2 \) and \( \delta_1 \leq \delta \leq \delta_2 \). The inferred probabilities will be distinguishable as high or low for sufficiently long emitted sequences.

Let \( d^+_h(u) \) and \( d^+_l(u) \) denote the number of outgoing arcs from \( u \) that are labelled high and low, respectively. We count each self-loop as a single arc.

**Lemma 4.** The sets \( \bar{U}_i \) and \( U_i \) for founder vertices can be constructed.

**Proof.** Suppose \( i \) is a founder vertex. Then from a state \( u \) in \( \bar{U}_i \), there are precisely \( m \) transitions with high probability. On the other hand, if \( i \) is not a founder vertex, then it has parents \( j \) and \( k \); therefore, from a state \( u \) in \( U_i \), there are at least \( |U_j \cup U_k| \geq m + 1 \) outgoing arcs that are labelled high. Observe also that if \( i \) a founder vertex, and \( u \) is in \( \bar{U}_i \) but not in \( \bar{U}_i \) then there will be at least \( m + 1 \) outgoing arcs from \( u \) that are labelled high, since \( u \) will also be in some other \( U_j \) in that case. Therefore, \( u \) is in \( \bar{U}_i \) for some founder vertex \( i \) if and only if \( d^+_h(u) = m \). The set of all such vertices in \( H \) naturally partitions into blocks, one block \( \bar{U}_i \) for each founder \( i \), since if \( \bar{U}_i \) and \( \bar{U}_j \) correspond to two founders, and \( u \in \bar{U}_i \) and \( v \in \bar{U}_j \) then there will be transitions from \( u \) to \( v \) and from \( v \) to \( u \) in the emitted sequence if and only if \( \bar{U}_i = \bar{U}_j \). Once \( \bar{U}_i \) is known for each founder \( i \), we can construct \( U_i \) as well: if there is an arc \( (u, v) \) that is labelled high for a state \( u \) in \( \bar{U}_i \).
and a state \( v \) not in \( \bar{U}_i \), where \( i \) is a founder vertex, then \( v \) must be in \( U_i \). □

In general, for vertices other than founders, we will be interested in constructing only \( \bar{U}_i \).

We treat the above construction as the base case of a recursive procedure for constructing all \( \bar{U}_i \).

Let \( \mathcal{F} = \{ \bar{U}_i \} \) be the collection that has been constructed so far. At the end of the base case, each \( \bar{U}_i; i \geq f \) is in \( \mathcal{F} \). The construction proceeds in a top-down manner; so if \( j \) and \( k \) are parents of \( i \), and if \( \bar{U}_i \) is in \( \mathcal{F} \), then \( \bar{U}_j \) and \( \bar{U}_k \) have already been constructed and added in \( \mathcal{F} \). Let \( \cup_S \) denote the union over all sets in \( \mathcal{F} \).

Let \( \bar{U}_j \) and \( \bar{U}_k \) be any two distinct sets in \( \mathcal{F} \) such that \( \bar{U}_i \) for children \( i \) with parents \( j \) and \( k \) have not been constructed so far.

Let \( T_{jk} \) be the set of states \( u \) for which the following conditions hold:

1. \( u \) is not in \( \cup_S \cup_{r \geq f} U_r \), and
2. there is a high arc \((u, w)\) in \( H \) for every \( w \) in \( \bar{U}_j \cup \bar{U}_k \)

Lemma 5. If a state \( u \) is in \( T_{jk} \) then it is in \( U_i \) for some child \( i \) with parents \( j \) and \( k \). If a state \( u \) is in \( \bar{U}_i \) for some child \( i \) with parents \( j \) and \( k \) then \( u \) is in \( T_{jk} \).

Proof. When the second condition holds it is possible that \( u \) is in \( U_j \cap U_k \) and both \( j \) and \( k \) are founders. But this possibility is eliminated by the first condition. Therefore \( u \) must be in \( U_i \) for some child \( i \) with parents \( j \) and \( k \). The second statement is then obvious. □

The above proposition implies that

\[ \cup_i \bar{U}_i \subseteq T_{jk} \subseteq \cup_i U_i, \]

where the unions are over the children of \( j \) and \( k \).

Lemma 6. Let \( u \) be a state in \( T_{jk} \). If \( u \) is in \( \bar{U}_i \) for some child \( i \) with parents \( j \) and \( k \) then \( d^+_h(u) = |U_j \cup U_k| \), (which may not be known). If \( u \) is not in \( U_i \) for any child \( i \) with parents \( j \) and \( k \), then \( d^+_h(u) \geq |U_j \cup U_k| + 1 \)

Proof. The first statement follows from the fact that \( u \) is not in any other set \( U_r \), and the second statement follows from the fact that \( u \) is in \( U_i \) for some child \( i \) with parents \( j \) and \( k \) and at least in one other \( U_r \). □

Corollary 1. The set \( \bar{T}_{jk} = \cup_i \bar{U}_i \), where the union is over children \( i \) of \( j \) and \( k \), is recognised.
Proof. The set $\bar{T}_{jk}$ is the set of states $u$ in $T_{jk}$ for which $d^h_u(u)$ is minimum. $\square$

Lemma 7. The set $\bar{T}_{jk}$ partitions into blocks $\bar{U}_i$ for children $i$ with parents $j$ and $k$.

Proof. States $v$ and $w$ in $T_{jk}$ are the same block if and only if there are arcs $(v, w)$ and $(w, v)$ labelled low. $\square$

This construction terminates when no more blocks can be added to $\mathcal{F}$, thus completing the proof of Theorem 1.

Remark 1. In the above construction we recognised $\bar{U}_i$ for all vertices in the pedigree. We also recognised the parent-child relationships between them, which allowed us to construct the whole pedigree on the single extant vertex. Now suppose that we have a pedigree on more than one extant individuals. For each extant vertex we have a sequence emitted by the automaton that corresponds to the sub-pedigree on that extant vertex. It is reasonable to suppose that each vertex $i$ in the pedigree corresponds to a unique $U_i \subset [N]$. Such a supposition means that the extant individuals that are descendents of $i$ (the cluster of $i$) share some common traits, and the states in $\bar{U}_i$ are observed only in the sequences of the extant individuals in the cluster of $i$. We, therefore, construct the pedigree of each extant individual separately. To construct a graph theoretic union of all these pedigrees, we identify vertices $y$ and $z$, respectively, in pedigrees $\mathcal{P}_i$ and $\mathcal{P}_j$ whenever $\bar{U}_y$ and $\bar{U}_z$ are identical. It is possible to generalise the correspondence between pedigrees and automata that was considered above to a correspondence between pedigrees on multiple extant vertices and more general automata in which there are transitions from a vertex either to its parents or to itself or to any of its extant descendents. The mechanism for emitting characters would not be essentially different. For example, when the automaton is in state $v$, (that is, at vertex $v$), it would emit characters from $U_v$ at all its descendents.

4.2. Example. We now illustrate the above construction with an example. The matrix $H$ below represents the directed graph $H$ that was defined earlier. Thus its vertex set is the set of states observed in the emitted sequence, which in our example is $\{1, 2, \ldots, 14\}$. The arcs of $H$ are labelled $h$ (high) or $l$ (low).
Observe that the rows 6, 7 and 8 have the minimum number 3 of \( h \), therefore, \( m = 3 \), and \( \cup_i \bar{U}_i = \{6, 7, 8\} \), where the union is over the indices of the founders. Also, observe the block structure of the sub-matrix consisting of rows and columns 6, 7 and 8: there are no arcs from 6 to 7 or 8, and no arcs from 7 or 8 to 6, but there are arcs between 7 and 8. Therefore, there are two founders in the pedigree.

There are outgoing arcs \((6, 10)\) and \((6, 11)\) that are labelled \( h \), therefore, the character set for one of the founders is \( U_f = \{6, 10, 11\} \). Similarly, the character set for the other founder is \( U_g = \{7, 8, 9\} \). We have called them \( U_f \) and \( U_g \) since we do not know how many vertices are in the pedigree; but the naming is not relevant. We now set \( F = \{\bar{U}_f = \{6\}, \bar{U}_g = \{7, 8\}\} \).

We now consider pairs \( \bar{U}_j \) and \( \bar{U}_k \) in \( S \). In this case there is only one pair. The matrix \( H \) shows 6 states 4,5,9,10,12,13 that have high-arcs to 6 and to \( \{7, 8\} \), and are therefore the candidate states for inclusion in \( \bar{U}_i \) for children of \( j \) and \( k \). We omit 10 from this list because 10 is in \( U_f \) but not in \( \bar{U}_f \). We then note that \( d_h^+(4) = d_h^+(5) = 6 \), while \( d_h^+(9) \), \( d_h^+(12) \), and \( d_h^+(13) \) are all more than 6. Therefore, we eliminate 9, 12 and 13 as well from the list of candidate states. Since there are no arcs between 4 and 5, the blocks to be included in \( F \) are \( \bar{U}_e = \{4\} \) and \( \bar{U}_d = \{5\} \). Both \( d \) and \( e \) are children of \( f \) and \( g \). Here we also conclude that since 9, 10, 11, 12 and 13 are in \( U_d \cup U_e \cup U_f \cup U_g \), they cannot be in any \( \bar{U}_i \) that will be discovered in future, so they do not have to be considered.
Next we have to repeat the process for all pairs of blocks in $F$ (except of course the ones which we have already processed in earlier steps).

Consider the pair $\bar{U}_e$ and $\bar{U}_g$. The states $2, 12, 13, 14$ have high-arcs to each state in $\bar{U}_e \cup \bar{U}_g = \{4, 7, 8\}$. But $12$ and $13$ have been eliminated before. Since $d^+_h(2) = d^+_h(2) = 6$, and there are arcs $(2, 14)$ and $(14, 2)$, there is only one new block $\bar{U}_c = \{2, 14\}$, and $c$ is a child of $e$ and $g$.

Next we claim that $d$ and $g$ have no child together since only state $13$ has high-arcs to all states in $\bar{U}_d \cup \bar{U}_g = \{5, 7, 8\}$, but $13$ has been eliminated earlier. By similar reasoning, we claim that vertices $e$ and $f$ do not have a child, and vertices $d$ and $f$ do not have a child.

Next we note that the states $3, 11$ and $13$ have high-arcs to all vertices in $\bar{U}_d \cup \bar{U}_e = \{4, 5\}$. But $11$ and $13$ were eliminated earlier. Therefore, the next block to be added to $F$ is $\bar{U}_b = \{3\}$.

Only $11$ and $13$ have high-arcs to all states in $U_f$ and $U_d$. But $11$ is in $U_f$, where $f$ is a founder, and $13$ has high-arcs to vertices in $\bar{U}_g$. Therefore, $d$ and $f$ have no children together.

In the end, we observe that the states $1, 9$, and $13$ have high-arcs to states in $\bar{U}_b \cup \bar{U}_c$, but $9$ and $13$ are discarded before, so we conclude the construction by adding block $\bar{U}_a = \{1\}$ to $F$, which corresponds to the extant vertex. The resulting pedigree is the one shown on the left of Figure 1.

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