Monotonic BART and outlier detection

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Chronically ill children and potential height outliers

- This data is from the electronic health records (EHR) of a large regional children's health care system
- Chronically ill children are often at high risk for malnutrition
- Typically this is assessed by comparison to US Centers for Disease Control (CDC) growth chart benchmarks (or WHO)
- CDC inputs are age, gender, height and weight
- Age and gender are extremely reliable
- However, height and weight are prone to EHR outliers and there is practically NO quality control for these measures i.e., the ground truth of height outliers is largely unknown
- Based on a UK study, there are an order of magnitude more height (3%) than weight outliers (0.2%) per measurement (Phan et al. 2020 Scientific Reports)
- Determining malnourishment requires height outlier detection
- Furthermore, this method should be robust to weight outliers that are harder to identify (yet fortunately less prevalent)

CDC Growth Charts: United States

Stature-for-age percentiles:
Girls, 2 to 20 years
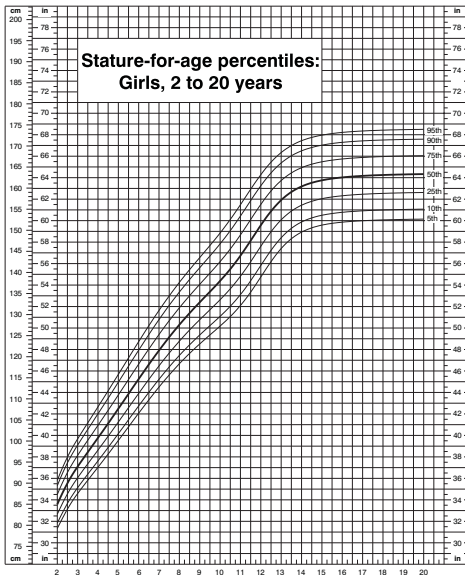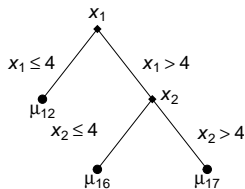
Monotonic BART (mBART)

Chipman et al. 2021 *Bayesian Analysis*

- $f \overset{\text{prior}}{\sim} \text{mBART}$

- A function f is monotone with respect to x_j if f satisfies
  $f(\dots, x_{j-1}, x_j + \Delta x, x_{j+1}, \dots) \geq f(\dots, x_{j-1}, x_j, x_{j+1}, \dots)$
  for all $\Delta x > 0$ (increasing/nondecreasing) or
  for all $\Delta x < 0$ (decreasing/nonincreasing)

- Constraint Conditions for Tree Monotonicity
  A tree function $g(x; \mathcal{T}, \mathcal{M})$ will be monotone in coordinate x_j
  if the leaf value of each of its terminal node regions is
  (a) not greater than the minimum level of all of its
  above-neighbor regions with respect to x_j and
  (b) not less than the maximum level of all of its
  below-neighbor regions with respect to x_j

Monotonic BART (mBART)

- ▶ dichotomous covariates are monotonic in the trivial sense
- ▶ for continuous outcomes, we can restrict to monotonicity
- ▶ e.g, assume age, t, is monotonic with respect to height, y
- ▶ do we want to assume the same for age, t, and weight, w?
- ▶ we can relax this restriction like so: $x = (t, u, v, w, -w)$
- ▶ yet from here on, we assume that weight is monotonic

Monotonic example: increasing in x_1 and x_2
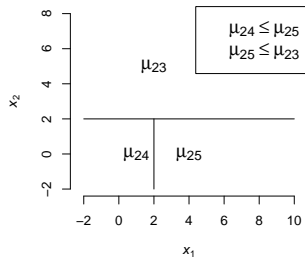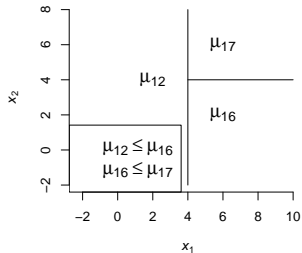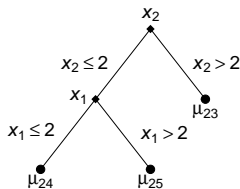
Monotonic BART (mBART)

Chipman et al. 2021 *Bayesian Analysis*

- The leaf prior for BART $\quad \mu_j | \mathcal{T} \overset{\text{prior}}{\sim} \mathrm{N}\big(0,\ \sigma_\mu^2\big)$
- Consider the simplest case of two monotonic leaves in mBART (relying on the results of Azzalini 1985 *Scand J Stat*)

$$
\left[\begin{array}{c} \mu_1 \\ \mu_2 \end{array} \right] \overset{\text{prior}}{\sim} \mathrm{N}_2 \big(\vec{0}_2,\ c^2\sigma_\mu^2 I_2\big)\, \mathrm{I}(\mu_1 < \mu_2) \ \text{where}\ c^2 = \frac{\pi}{\pi-1} \approx 1.47
$$

Equivalent to skew Normal marginals where $\mathrm{V}\,[\mu_1] = \mathrm{V}\,[\mu_2] = \sigma_\mu^2$

$$
\mu_1 \overset{\text{prior}}{\sim} \phi\left(\frac{\mu_1}{c\,\sigma_\mu}\right) \Phi\left(\frac{-\mu_1}{c\,\sigma_\mu}\right) \qquad \mathrm{E}\,[\mu_1] = \frac{-\sigma_\mu}{\sqrt{\pi-1}}
$$

$$
\mu_2 \overset{\text{prior}}{\sim} \phi\left(\frac{\mu_2}{c\,\sigma_\mu}\right) \Phi\left(\frac{\mu_2}{c\,\sigma_\mu}\right) \qquad \mathrm{E}\,[\mu_2] = \frac{+\sigma_\mu}{\sqrt{\pi-1}}
$$

Nonparametric outlier detection

- *Monotonicity provides additional robustness to outliers since f can't just rise towards an outlier and double-back down afterward (nor in the inverse directions)*
- We have *population* predictions of the form $\hat{y}_{ij} = \mathrm{E}\left[y_{ij}\right] = \hat{f}(x_{ij})$ where $j = 1, \ldots, n_i$
- *But these expectations are biased except for the average child*
- We need to adjust these up or down for a given subject
- So let $m_i = \mathrm{median}_j(y_{ij} - \hat{y}_{ij})$ (median rather than mean to be robust to outliers)
- Now, we make *personalized* predictions $\tilde{y}_{ij} = m_i + \hat{y}_{ij}$
- We define the *relative error* of these as $d_{ij} = (y_{ij} - \tilde{y}_{ij})/\tilde{y}_{ij}$
- Outliers are defined as $|d_{ij}| > \delta$ where δ can be determined from the Receiver Operating Characteristic (ROC) curve
- And the discriminating performance of the method is assessed by the area under the ROC curve

Friedman's partial dependence function and the Synthetic Approximation marginal

- t for age, u for sex, v for race and w for weight
- To do this right, first consider the likely monotonic relationship between age, sex and weight
  $$\mathrm{E}\,[w|t,u] = \tilde{f}(t,u)$$
- So we need to fit an intermediate mBART model first
  $$w_i = \tilde{f}(t_i, u_i) + \tilde{\epsilon}_i \text{ where } \tilde{f} \overset{\mathrm{prior}}{\sim} \mathrm{mBART}$$
- The Synthetic Approximation marginal for dependent variables

$$f_{t,u}(t,u) = \mathrm{E}_v\left[f(t,u,v,w^*)|t,u,w^* = \mathrm{E}[w|t,u]\right]$$
$$= \mathrm{E}_v\left[f(t,u,v,\tilde{f}(t,u))|t,u\right]$$

Returning to the real data example

Sparapani et al. 2022 *JPGN*

- ▶ Constructed two independent cohorts of chronically ill children
  - ▶ 2-8 years old
  - ▶ measured at least every 120 days on average
  - ▶ followed for at least 2 years
- ▶ Training cohort: 1376 children with height outliers unknown
  39491 measurements: 28.7/child on average
- ▶ Validation cohort: 318 children
  7378 measurements: 23.2/child on average
  manually reviewed to determine height outliers
  however, the *ground truth* is fallible
  it is retrospective: we can't just re-measure these children
- ▶ Heights in the training cohort fit with mBART to
  age, sex, race/ethnicity and weight

Returning to the real data example

- Outlier detection conducted for the Validation cohort
- The area under the Receiver Operating Characteristic (ROC) curve was excellent: 0.841
- By comparison, if you use the CDC height by age growth chart, the area is only 0.776
- Based on ROC curve, two relative error cutoffs considered Aggressive, 0.075; and Conservative, 0.085

Real data summary

	Training		Validation	
	1376		318	
Children	n	(%)	n	(%)
Female	594	(43.2%)	132	(41.5%)
Black	313	(22.7%)	66	(20.8%)
White	783	(56.9%)	189	(59.4%)
Other	280	(20.3%)	63	(19.8%)
Children with outliers	N/A		101	(31.8%)
Measurements	m		m	
Height (cm)	39491		7378	
	Mean	(SD)	Mean	(SD)
Measurements/child	28.7		23.2	
First visit age 2	86.4	(8.8)	84.5	(6.6)
Last visit age 5	111.3	(8.4)	109.5	(9.4)
	%		%	
mBART R^2	82.2%		75.3%	

FPD marginal of age: assuming Independence
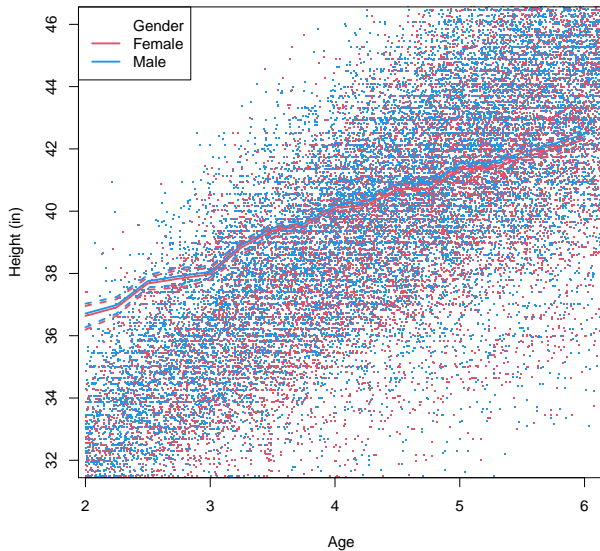
SA marginal of age: assuming Dependence
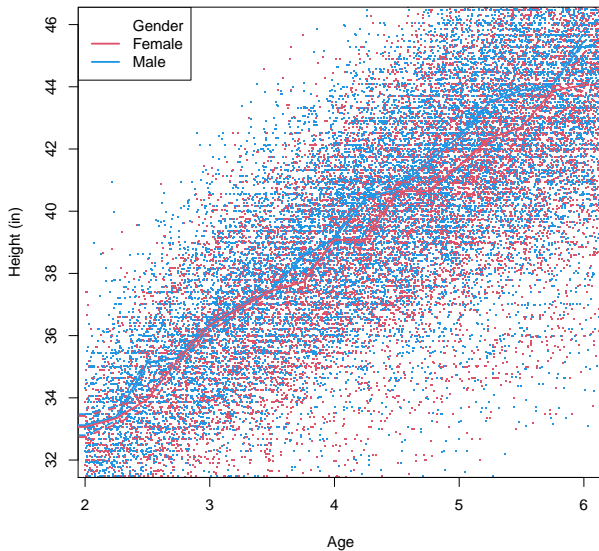
FPD marginal of weight: assuming Independence
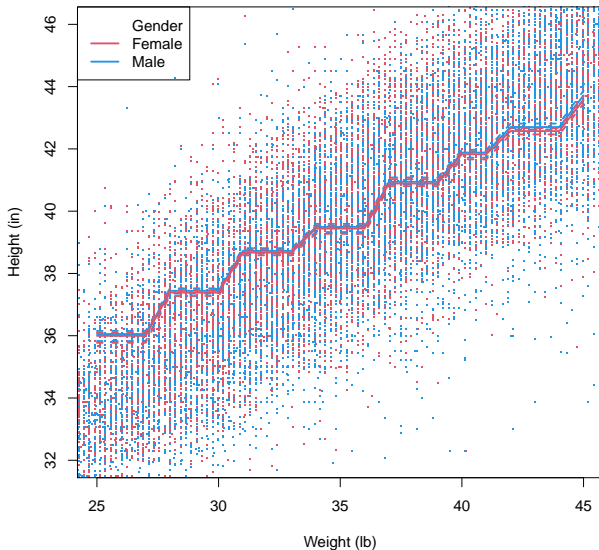
SA marginal of weight: assuming Dependence
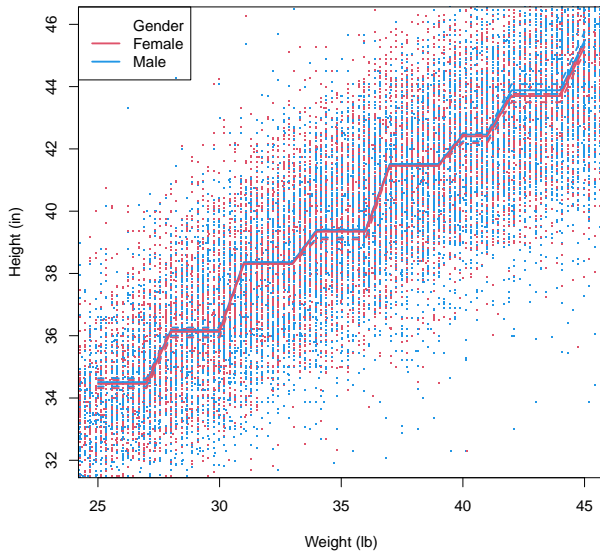
Receiver Operating Characteristic (ROC) curve
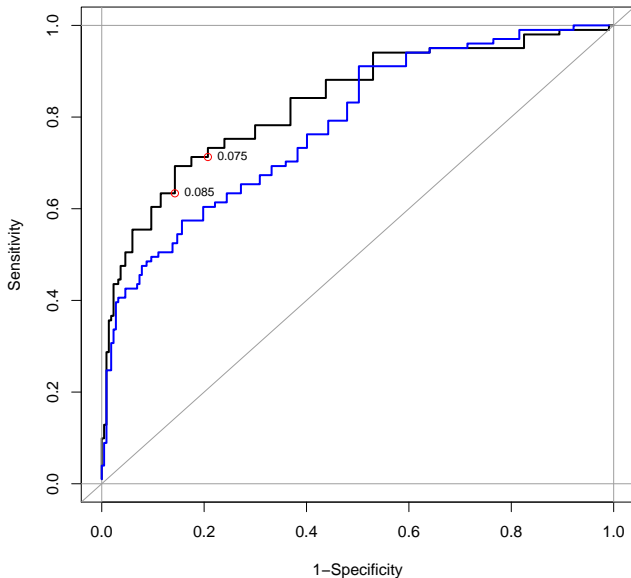
Aggressive cutoff 0.075

- ▶ B: mBART outlier detection
- ▶ C: clinical review ground truth
- ▶ Outlier: 0 (False), 1 (True)

|	B=0	B=1	
C=0	TN=172	FP=45	M=217
C=1	FN=29	TP=72	Q=101
	N=201	P=117	T=318

$$\text{Sensitivity or Recall} = P[B = 1 | C = 1] = \frac{TP}{Q} = \frac{72}{101} = 0.713$$

$$\text{Specificity} = P[B = 0 | C = 0] = \frac{TN}{M} = \frac{172}{217} = 0.793$$

$$\text{PPV or Precision} = P[C = 1 | B = 1] = \frac{TP}{P} = \frac{72}{117} = 0.615$$

$$\text{NPV} = P[C = 0 | B = 0] = \frac{TN}{N} = \frac{172}{201} = 0.856$$

Conservative cutoff 0.085

- ▶ B: mBART outlier detection
- ▶ C: clinical review ground truth
- ▶ Outlier: 0 (False), 1 (True)

	B=0	B=1	
C=0	TN=186	FP=31	M=217
C=1	FN=37	TP=64	Q=101
	N=223	P=95	T=318

$$\text{Sensitivity or Recall} = \mathbf{P}[B = 1 | C = 1] = \frac{TP}{Q} = \frac{64}{101} = 0.634$$

$$\text{Specificity} = \mathbf{P}[B = 0 | C = 0] = \frac{TN}{M} = \frac{186}{217} = 0.857$$

$$\text{PPV or Precision} = \mathbf{P}[C = 1 | B = 1] = \frac{TP}{P} = \frac{64}{95} = 0.674$$

$$\text{NPV} = \mathbf{P}[C = 0 | B = 0] = \frac{TN}{N} = \frac{186}{223} = 0.834$$

Aggressive cutoff: targeted smoothing BART with monotonic weight

Starling et al. Annals of Applied Statistics 2020

- ▶ B: mBART outlier detection
- ▶ C: clinical review ground truth
- ▶ Outlier: 0 (False), 1 (True)

|	B=0	B=1	
C=0	TN=165	FP=52	M=217
C=1	FN=27	TP=74	Q=101
	N=192	P=126	T=318

$$\text{Sensitivity or Recall} = P[B = 1 | C = 1] = \frac{TP}{Q} = \frac{74}{101} = 0.732$$

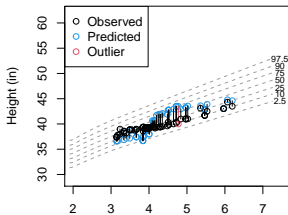$$\text{Specificity} = P[B = 0 | C = 0] = \frac{TN}{M} = \frac{165}{217} = 0.760$$

$$\text{PPV or Precision} = P[C = 1 | B = 1] = \frac{TP}{P} = \frac{74}{126} = 0.587$$

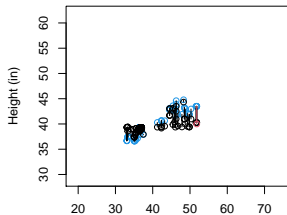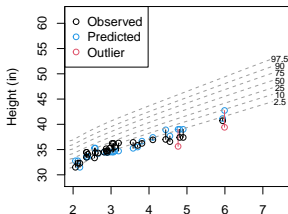$$\text{NPV} = P[C = 0 | B = 0] = \frac{TN}{N} = \frac{165}{192} = 0.859$$

Aggressive cutoff: females only

- B: mBART outlier detection
- C: clinical review ground truth
- Outlier: 0 (False), 1 (True)

	B=0	B=1	
C=0	TN=70	FP=20	M=90
C=1	FN=11	TP=31	Q=42
	N=81	P=51	T=132

$$\text{Sensitivity or Recall} = P[B = 1|C = 1] = \frac{TP}{Q} = \frac{31}{42} = 0.738$$

$$\text{Specificity} = P[B = 0|C = 0] = \frac{TN}{M} = \frac{70}{90} = 0.778$$

$$\text{PPV or Precision} = P[C = 1|B = 1] = \frac{TP}{P} = \frac{31}{51} = 0.608$$

$$\text{NPV} = P[C = 0|B = 0] = \frac{TN}{N} = \frac{70}{81} = 0.864$$

Aggressive cutoff: non-whites only

- ▶ B: mBART outlier detection
- ▶ C: clinical review ground truth
- ▶ Outlier: 0 (False), 1 (True)

	B=0	B=1	
C=0	TN=60	FP=19	M=79
C=1	FN=11	TP=39	Q=50
	N=71	P=58	T=129

$$\text{Sensitivity or Recall} = P[B = 1|C = 1] = \frac{TP}{Q} = \frac{39}{50} = 0.780$$

$$\text{Specificity} = P[B = 0|C = 0] = \frac{TN}{M} = \frac{60}{79} = 0.759$$

$$\text{PPV or Precision} = P[C = 1|B = 1] = \frac{TP}{P} = \frac{39}{58} = 0.672$$

$$\text{NPV} = P[C = 0|B = 0] = \frac{TN}{N} = \frac{60}{71} = 0.845$$

True Positives

(a) Sex F, Race W

(b) Sex F, Race W

(c) Sex F, Race B
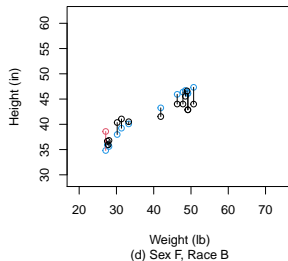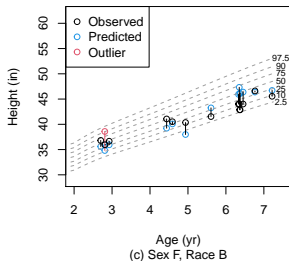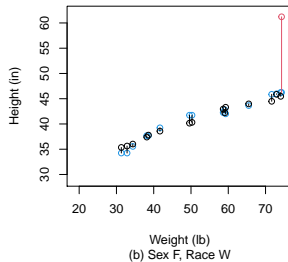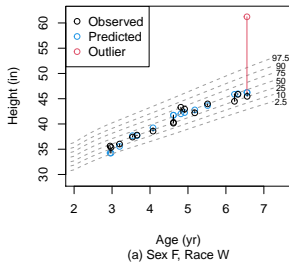
(d) Sex F, Race B

False Positives

(a) Sex M, Race W

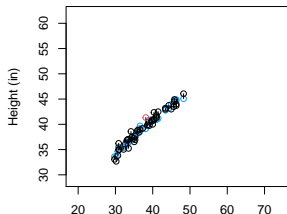(b) Sex M, Race W

(c) Sex M, Race B

(d) Sex M, Race B

False Negatives
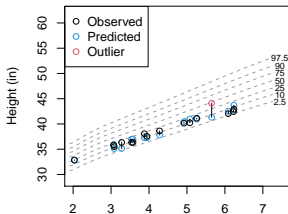
(a) Sex F, Race W
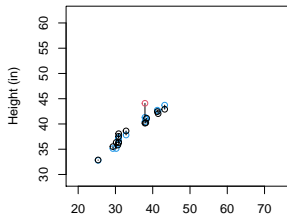
(b) Sex F, Race W
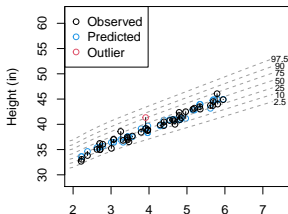
(c) Sex M, Race W

(d) Sex M, Race W

Conclusions

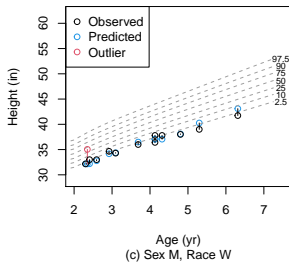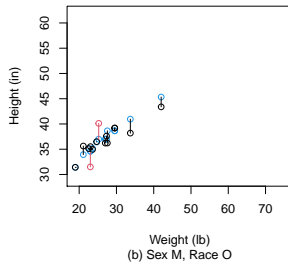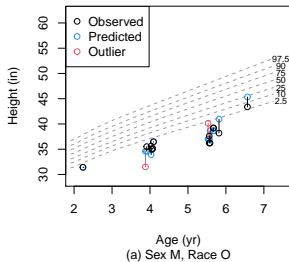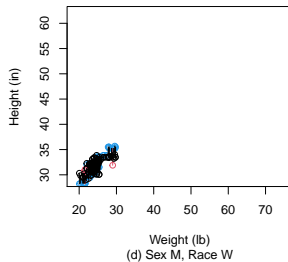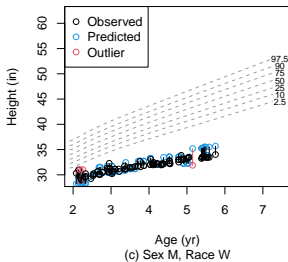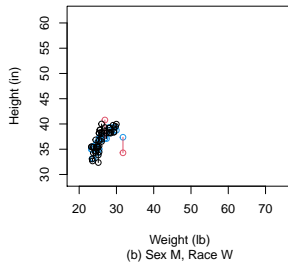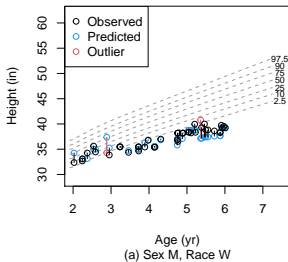- We constructed our new outlier detection methodology based on nonparametric machine learning with monotonic BART

- This automated method's performance was deemed to be adequate via an indpendent validation cohort

- Modern methodology leads to a simply-tuned single rule as opposed to complex simultaneous tuning of multiple rules that have been proposed (by others) based on classic methods

- For EHR heights/weights, the ground truth is unknown prospective corrections are rarely performed and retrospective attempts to identify outliers manually are fallible

- For covariates that have dependent relationships, we provide an extension to compute the marginal effects for one, or more, of these variables at a time: this extension has wide applicability in nonparametric/machine learning regression

True Positives

False Positives

(a) Sex M, Race W

(b) Sex M, Race W

(c) Sex M, Race W

(d) Sex M, Race W

False Negatives

(a) Sex M, Race B

(b) Sex M, Race B

(c) Sex F, Race W

(d) Sex F, Race W