

# Summary Gen4R

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## Terminology

- three\_present\_three
  - Three variants present and all three samples have matching GT
- three\_present\_two
  - Three variants present and two samples have matching GT
- three\_present\_zero
  - Three variants present and zero samples have matching GT
- two\_present\_two
  - Two variants present and two samples have matching GT

- two\_present\_zero
  - Two variants present and zero samples have matching GT
- one\_present
  - One variant present

## Sample Layout

- HG002
  - HM24385: LCL
  - HM26105: iPSC from LCL
  - HM27730: iPSC from PBMC
- HG004
  - HM24143: LCL
  - HM26077: iPSC from LCL
- HG005
  - HM24631: LCL
  - HM26107: iPSC from LCL
- Fibroblast
  - HM20431: LCL from Fibroblast
  - HM23248\_2:
  - HM23338: iPSC from Fibroblast

## Compare HG002: HM24385, HM26105, HM27730

- HM24385: LCL
- HM26105: iPSC from LCL
- HM27730: iPSC from PBMC

## Depth stats

### Depth quantiles (per match status)

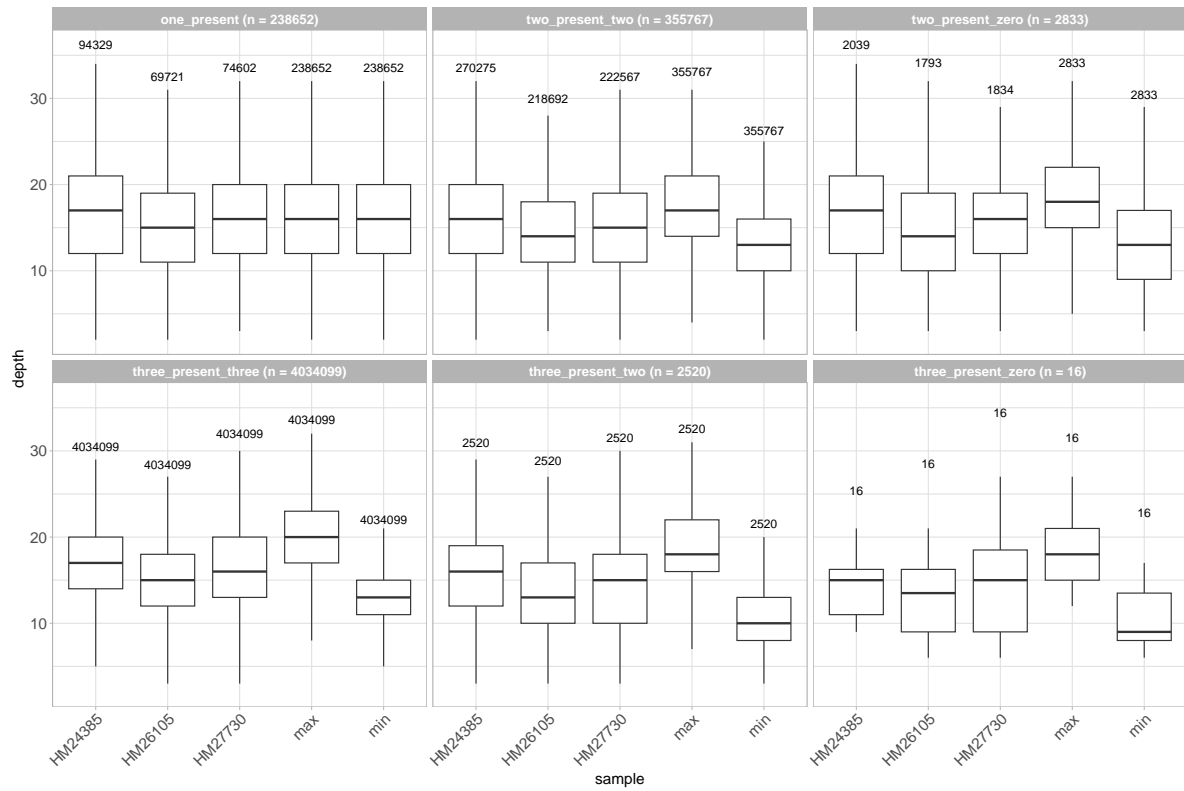
id	n	% total	0%	25%	50%	75%	100%
one_present	238652	5.15	2	12	16.0	20	588

three_present_three	4034099	87.06	2	13	16.0	20	598
three_present_two	2520	0.05	3	10	15.0	19	129
three_present_zero	16	0.00	6	9	14.5	17	27
two_present_two	355767	7.68	2	11	15.0	19	595
two_present_zero	2833	0.06	3	11	16.0	20	159
total	4633887	—	—	—	—	—	—

### Depth quantiles (per match/sample status)

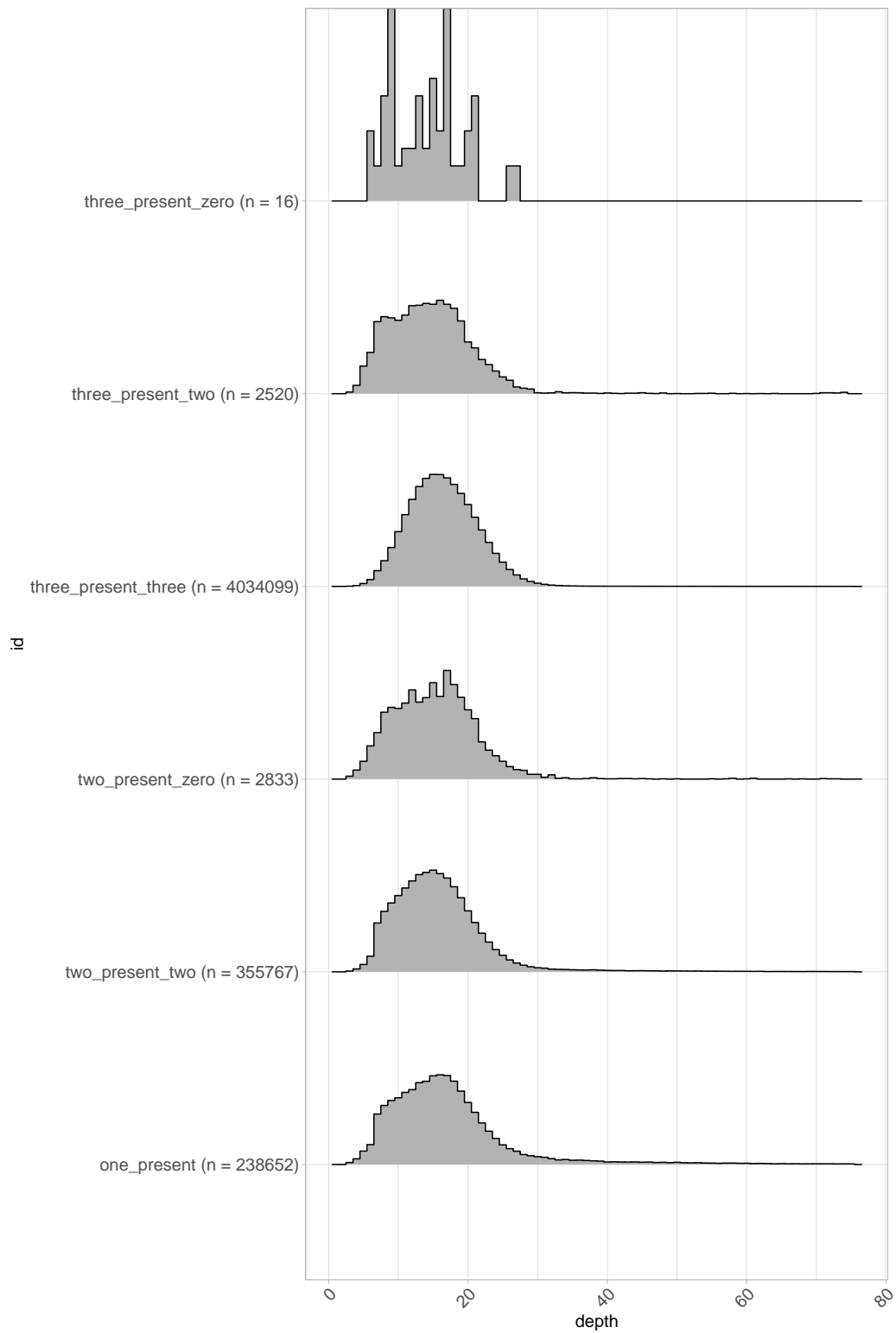
id	sample	n	% sample	0%	25%	50%	75%	100%
one_present	HM24385	94329	2.14	2	12	17.0	21.00	588
one_present	HM26105	69721	1.61	2	11	15.0	19.00	574
one_present	HM27730	74602	1.72	3	12	16.0	20.00	587
one_present	max	238652	5.15	2	12	16.0	20.00	588
one_present	min	238652	5.15	2	12	16.0	20.00	588
three_present_three	HM24385	4034099	91.62	2	14	17.0	20.00	598
three_present_three	HM26105	4034099	93.23	2	12	15.0	18.00	594
three_present_three	HM27730	4034099	93.05	2	13	16.0	20.00	593
three_present_three	max	4034099	87.06	3	17	20.0	23.00	598
three_present_three	min	4034099	87.06	2	11	13.0	15.00	593
three_present_two	HM24385	2520	0.06	3	12	16.0	19.00	129
three_present_two	HM26105	2520	0.06	3	10	13.0	17.00	101
three_present_two	HM27730	2520	0.06	3	10	15.0	18.00	111
three_present_two	max	2520	0.05	6	16	18.0	22.00	129
three_present_two	min	2520	0.05	3	8	10.0	13.00	101
three_present_zero	HM24385	16	0.00	9	11	15.0	16.25	21
three_present_zero	HM26105	16	0.00	6	9	13.5	16.25	21
three_present_zero	HM27730	16	0.00	6	9	15.0	18.50	27
three_present_zero	max	16	0.00	12	15	18.0	21.00	27
three_present_zero	min	16	0.00	6	8	9.0	13.50	17
two_present_two	HM24385	270275	6.14	2	12	16.0	20.00	595
two_present_two	HM26105	218692	5.05	3	11	14.0	18.00	593
two_present_two	HM27730	222567	5.13	2	11	15.0	19.00	593
two_present_two	max	355767	7.68	3	14	17.0	21.00	595
two_present_two	min	355767	7.68	2	10	13.0	16.00	593
two_present_zero	HM24385	2039	0.05	3	12	17.0	21.00	129
two_present_zero	HM26105	1793	0.04	3	10	14.0	19.00	159
two_present_zero	HM27730	1834	0.04	3	12	16.0	19.00	125
two_present_zero	max	2833	0.06	4	15	18.0	22.00	159
two_present_zero	min	2833	0.06	3	9	13.0	17.00	116

## Depth Boxplots





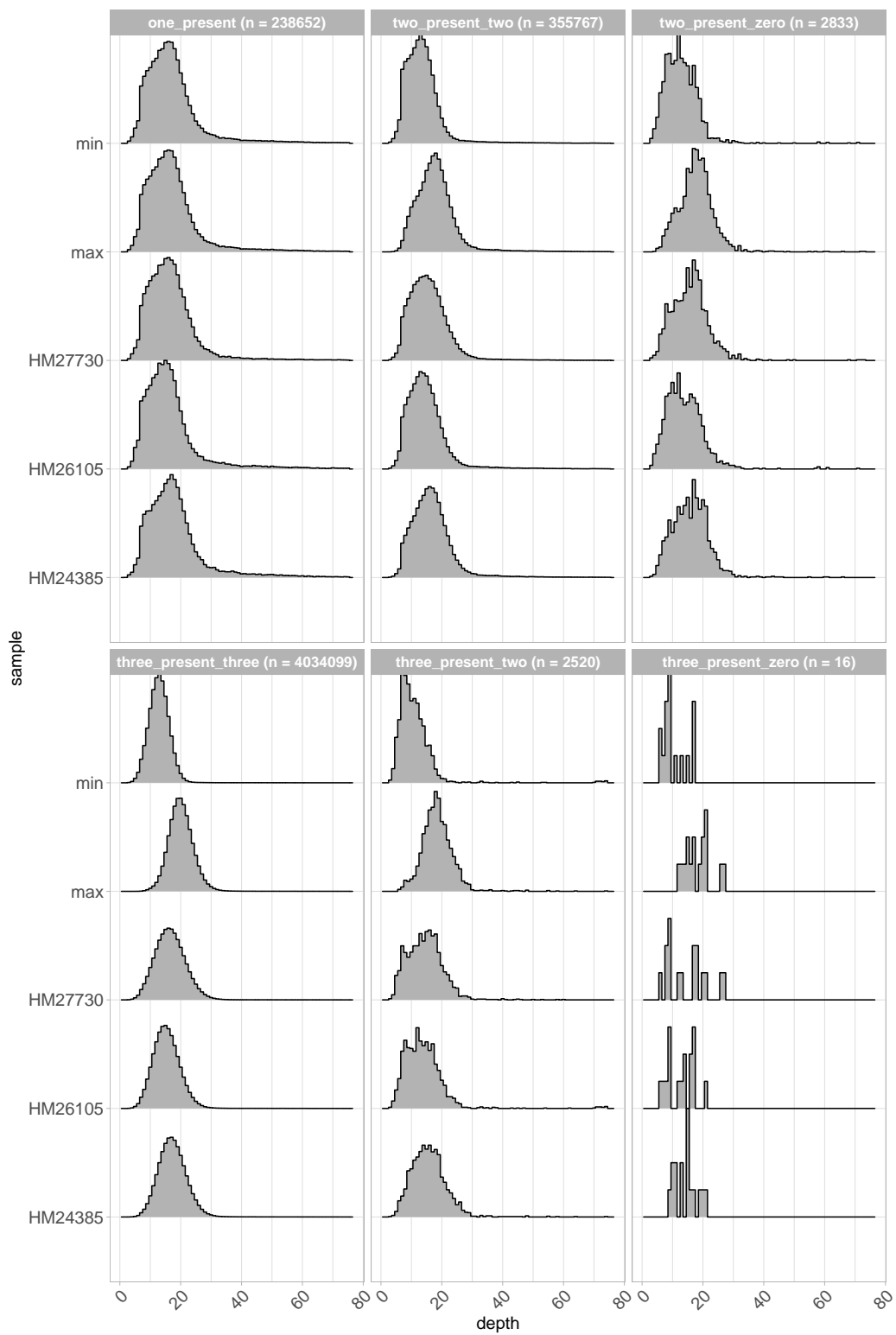
## Depth Histograms (per match status)







## Depth Histograms (per match/sample status)



## GQ stats

### GQ quantiles (per match status)

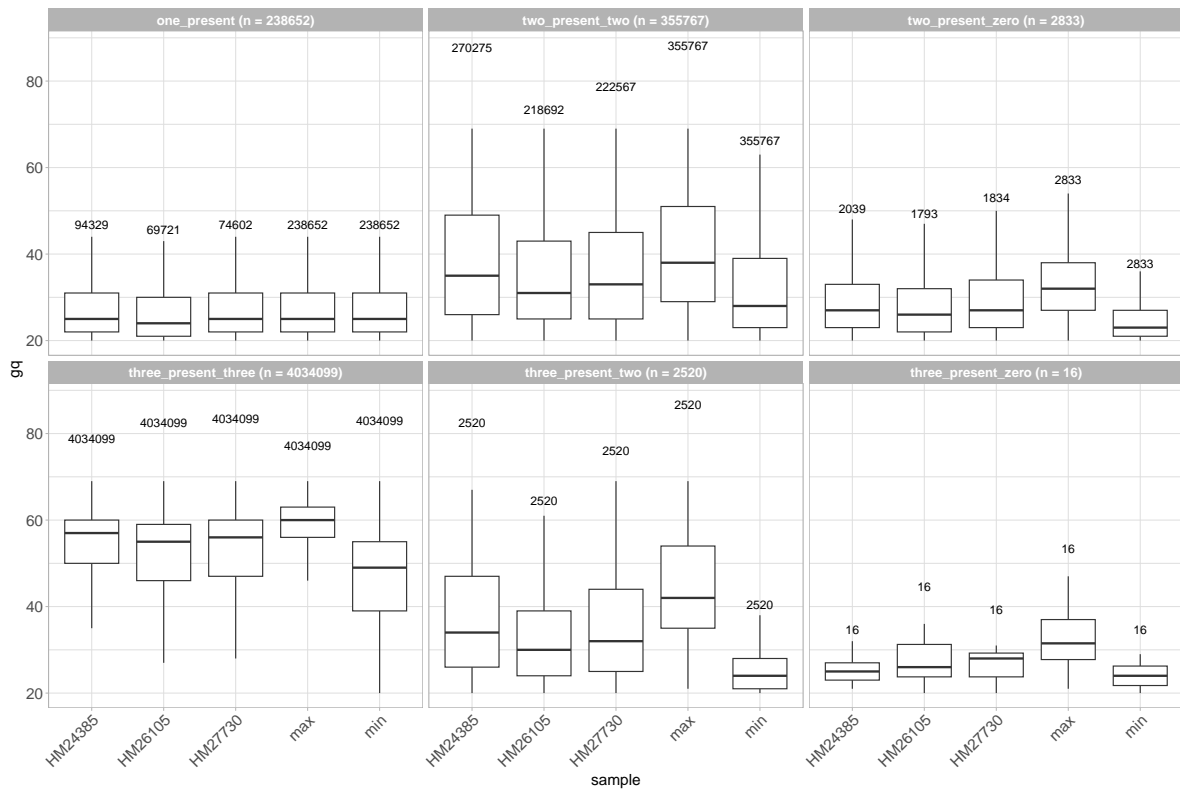
id	n	% total	0%	25%	50%	75%	100%
one_present	238652	5.15	20	22	25.0	31	67
three_present_three	4034099	87.06	20	47	56.0	60	69
three_present_two	2520	0.05	20	24	32.0	43	69
three_present_zero	16	0.00	20	23	26.5	31	47
two_present_two	355767	7.68	20	25	33.0	46	69
two_present_zero	2833	0.06	20	23	27.0	33	67
total	4633887	—	—	—	—	—	—

### GQ quantiles (per match/sample status)

id	sample	n	% sample	0%	25%	50%	75%	100%
one_present	HM24385	94329	2.14	20	22.00	25.0	31.00	67
one_present	HM26105	69721	1.61	20	21.00	24.0	30.00	67
one_present	HM27730	74602	1.72	20	22.00	25.0	31.00	67
one_present	max	238652	5.15	20	22.00	25.0	31.00	67
one_present	min	238652	5.15	20	22.00	25.0	31.00	67
three_present_three	HM24385	4034099	91.62	20	50.00	57.0	60.00	69
three_present_three	HM26105	4034099	93.23	20	46.00	55.0	59.00	69
three_present_three	HM27730	4034099	93.05	20	47.00	56.0	60.00	69
three_present_three	max	4034099	87.06	20	56.00	60.0	63.00	69
three_present_three	min	4034099	87.06	20	39.00	49.0	55.00	69
three_present_two	HM24385	2520	0.06	20	26.00	34.0	47.00	67
three_present_two	HM26105	2520	0.06	20	24.00	30.0	39.00	67
three_present_two	HM27730	2520	0.06	20	25.00	32.0	44.00	69
three_present_two	max	2520	0.05	21	35.00	42.0	54.00	69
three_present_two	min	2520	0.05	20	21.00	24.0	28.00	59
three_present_zero	HM24385	16	0.00	21	23.00	25.0	27.00	47
three_present_zero	HM26105	16	0.00	20	23.75	26.0	31.25	36
three_present_zero	HM27730	16	0.00	20	23.75	28.0	29.25	46
three_present_zero	max	16	0.00	21	27.75	31.5	37.00	47
three_present_zero	min	16	0.00	20	21.75	24.0	26.25	29
two_present_two	HM24385	270275	6.14	20	26.00	35.0	49.00	69
two_present_two	HM26105	218692	5.05	20	25.00	31.0	43.00	69
two_present_two	HM27730	222567	5.13	20	25.00	33.0	45.00	69
two_present_two	max	355767	7.68	20	29.00	38.0	51.00	69

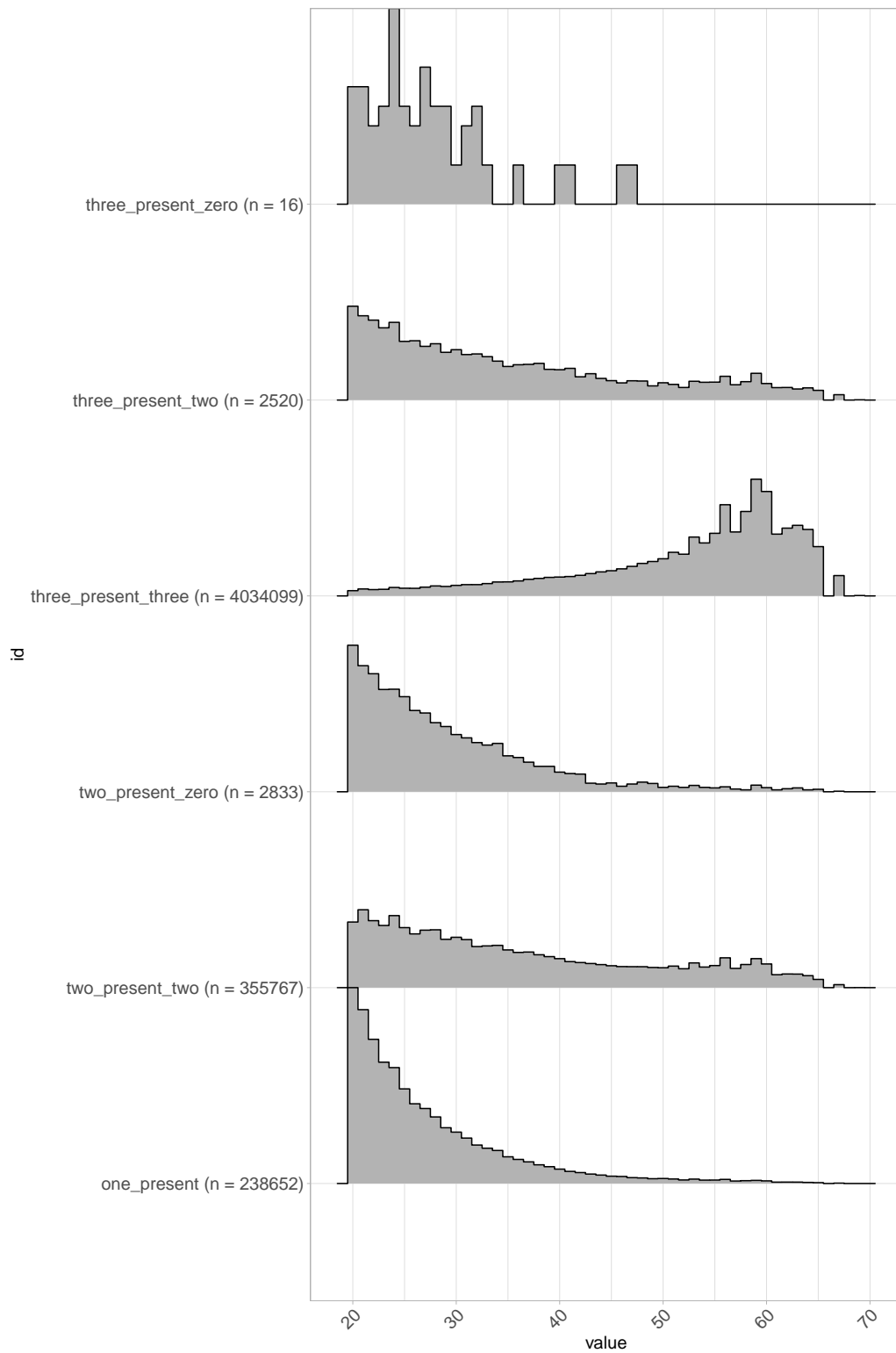
two_present_two	min	355767	7.68	20	23.00	28.0	39.00	67
two_present_zero	HM24385	2039	0.05	20	23.00	27.0	33.00	63
two_present_zero	HM26105	1793	0.04	20	22.00	26.0	32.00	61
two_present_zero	HM27730	1834	0.04	20	23.00	27.0	34.00	67
two_present_zero	max	2833	0.06	20	27.00	32.0	38.00	67
two_present_zero	min	2833	0.06	20	21.00	23.0	27.00	54

## GQ Boxplots



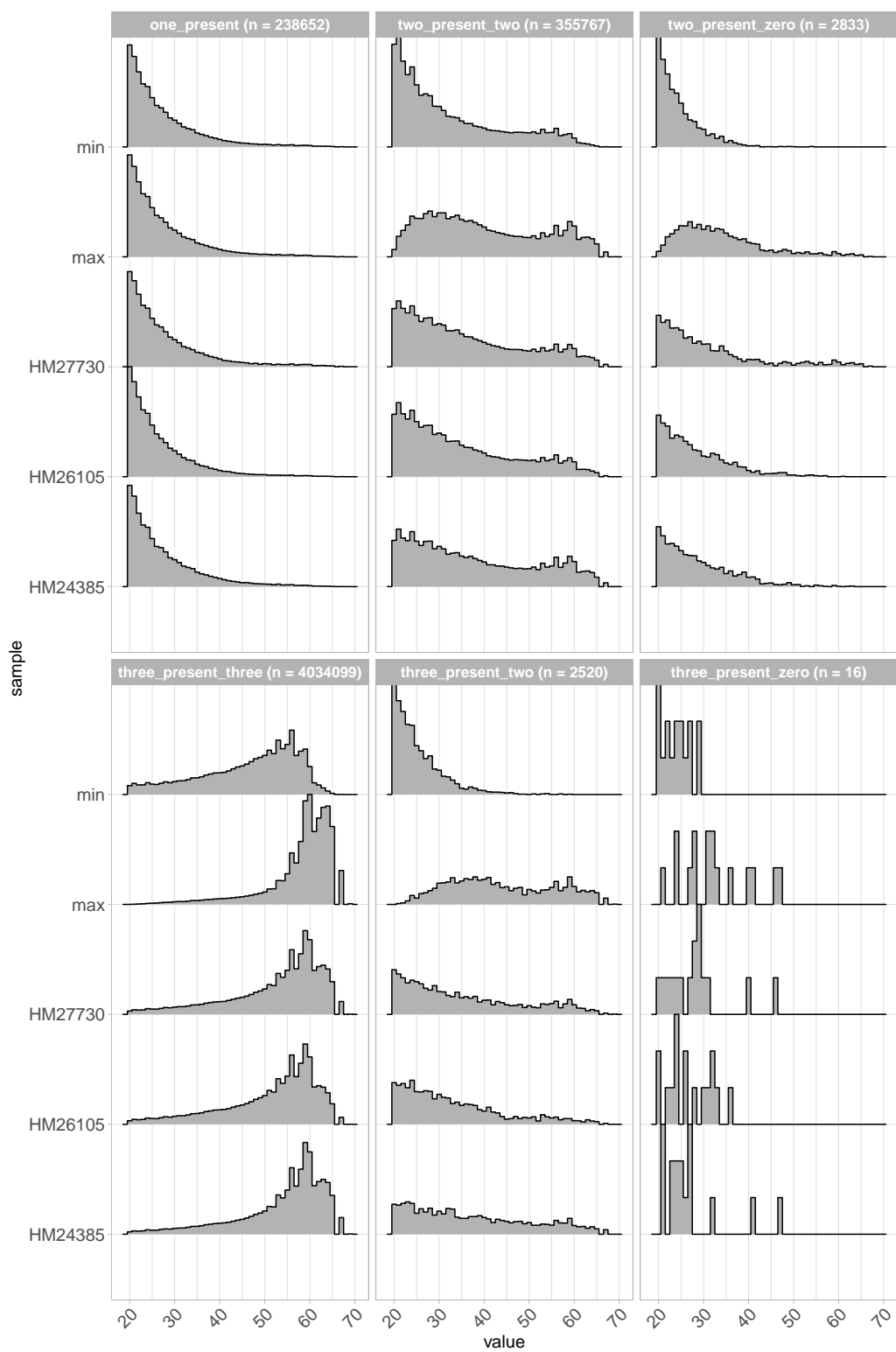


## GQ Histograms (per match status)





## GQ Histograms (per match/sample status)



## Compare HG004: HM24143, HM26077

- HM24143: LCL
- HM26077: iPSC from LCL

### Depth stats

#### Depth quantiles (per match status)

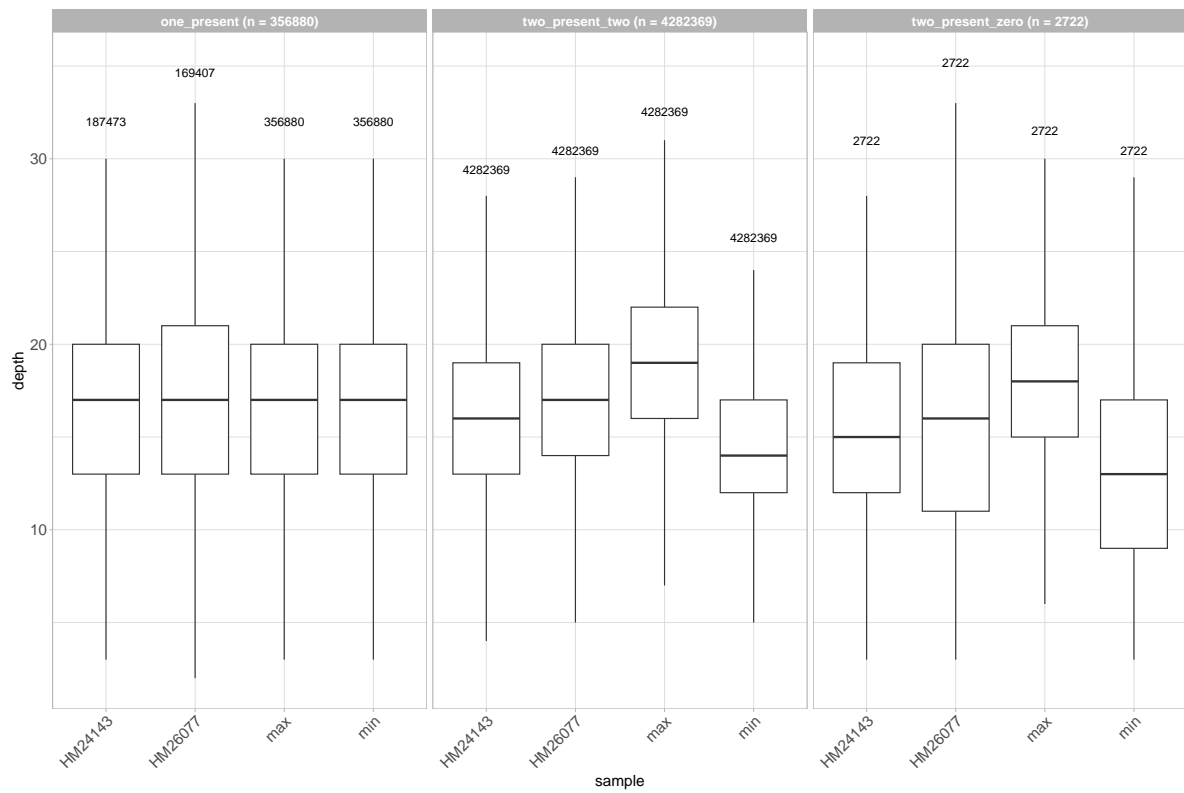
id	n	% total	0%	25%	50%	75%	100%
one_present	356880	7.69	2	13	17	20	594
two_present_two	4282369	92.25	2	14	16	20	595
two_present_zero	2722	0.06	3	12	16	20	164
total	4641971	—	—	—	—	—	—

#### Depth quantiles (per match/sample status)

id	sample	n	% sample	0%	25%	50%	75%	100%
one_present	HM24143	187473	4.19	2	13	17	20	592
one_present	HM26077	169407	3.80	2	13	17	21	594
one_present	max	356880	7.69	2	13	17	20	594
one_present	min	356880	7.69	2	13	17	20	594
two_present_two	HM24143	4282369	95.75	2	13	16	19	595
two_present_two	HM26077	4282369	96.14	2	14	17	20	595
two_present_two	max	4282369	92.25	3	16	19	22	595
two_present_two	min	4282369	92.25	2	12	14	17	593
two_present_zero	HM24143	2722	0.06	3	12	15	19	164
two_present_zero	HM26077	2722	0.06	3	11	16	20	164
two_present_zero	max	2722	0.06	5	15	18	21	164
two_present_zero	min	2722	0.06	3	9	13	17	140

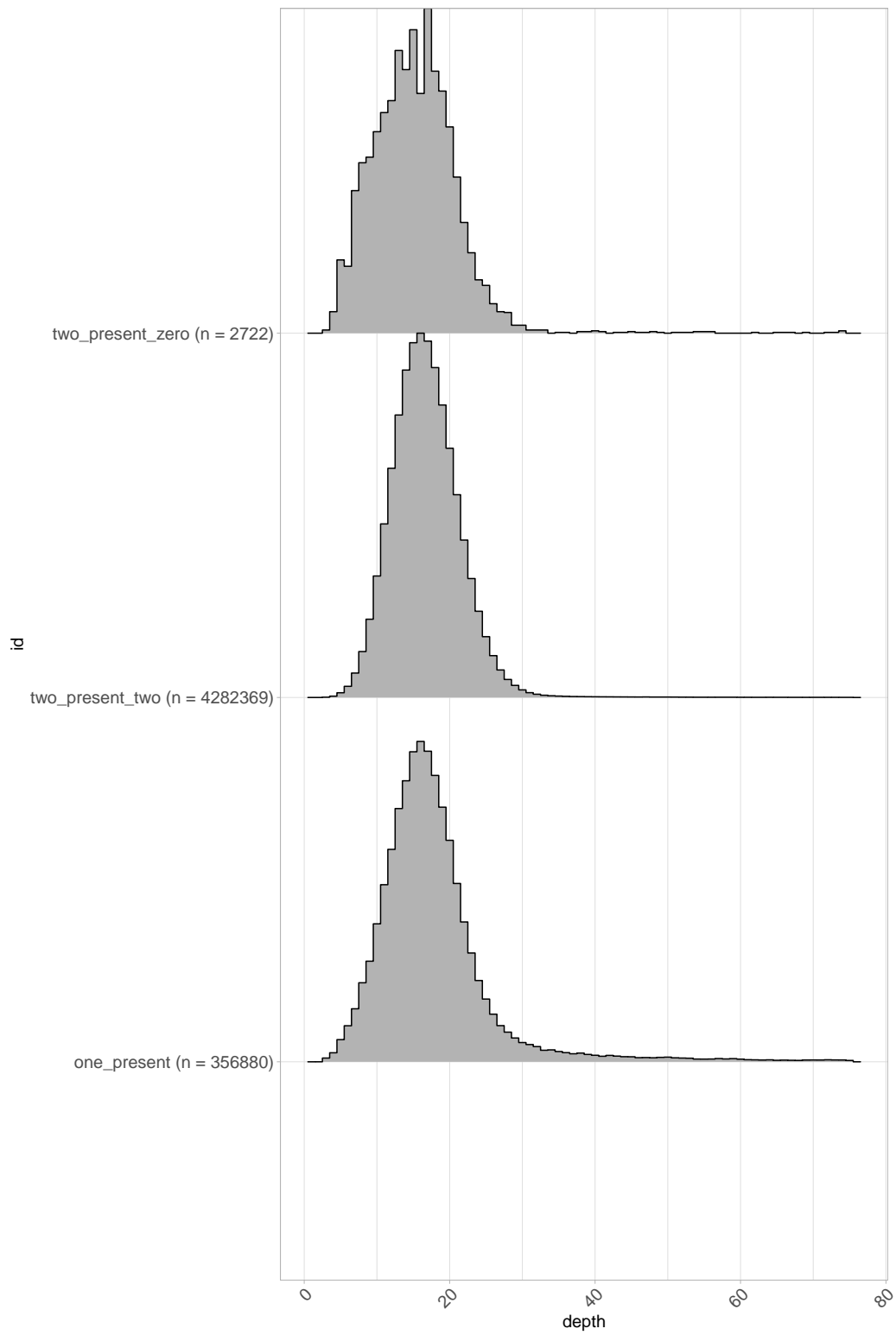


## Depth Boxplots



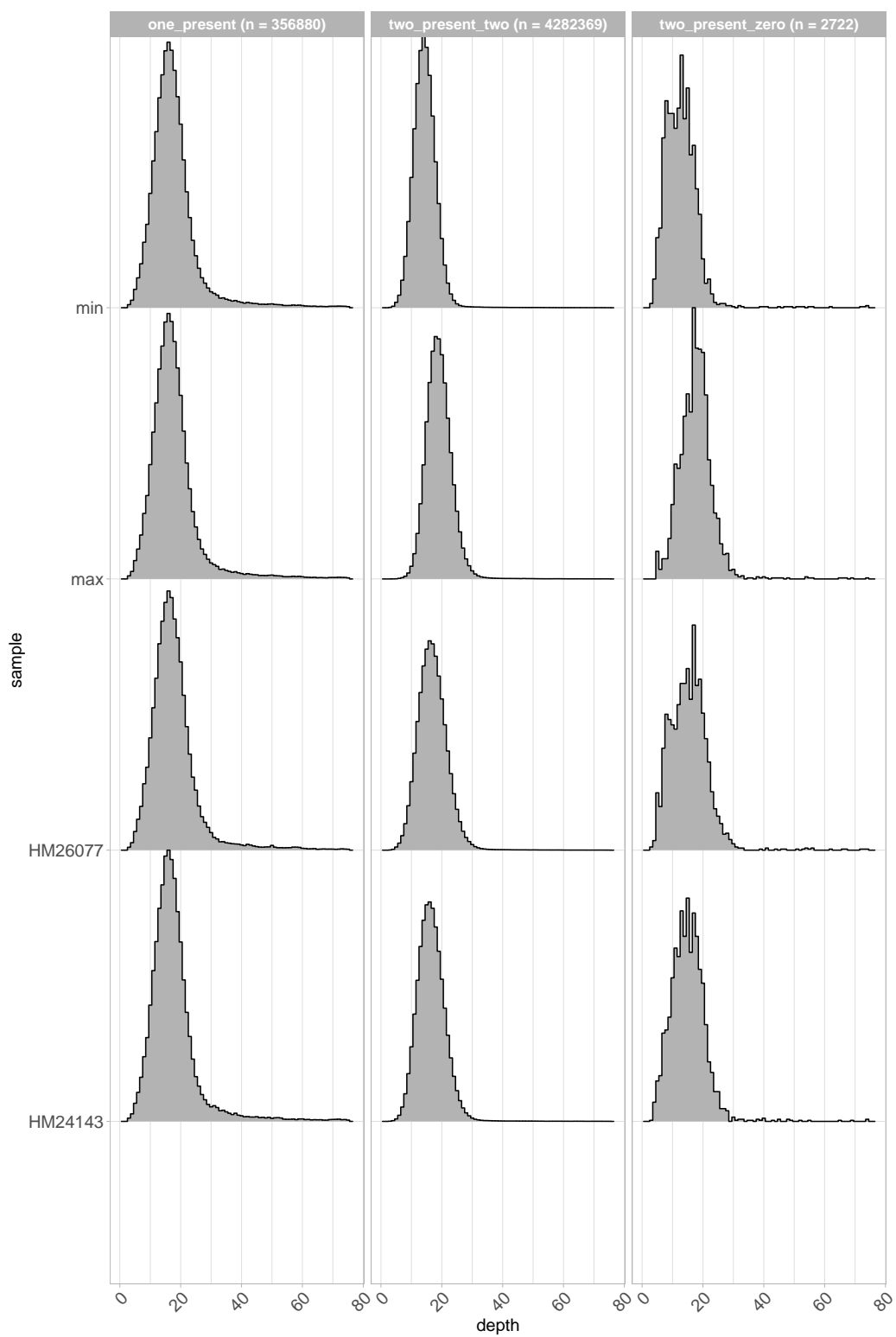


## Depth Histograms (per match status)





## Depth Histograms (per match/sample status)



## GQ stats

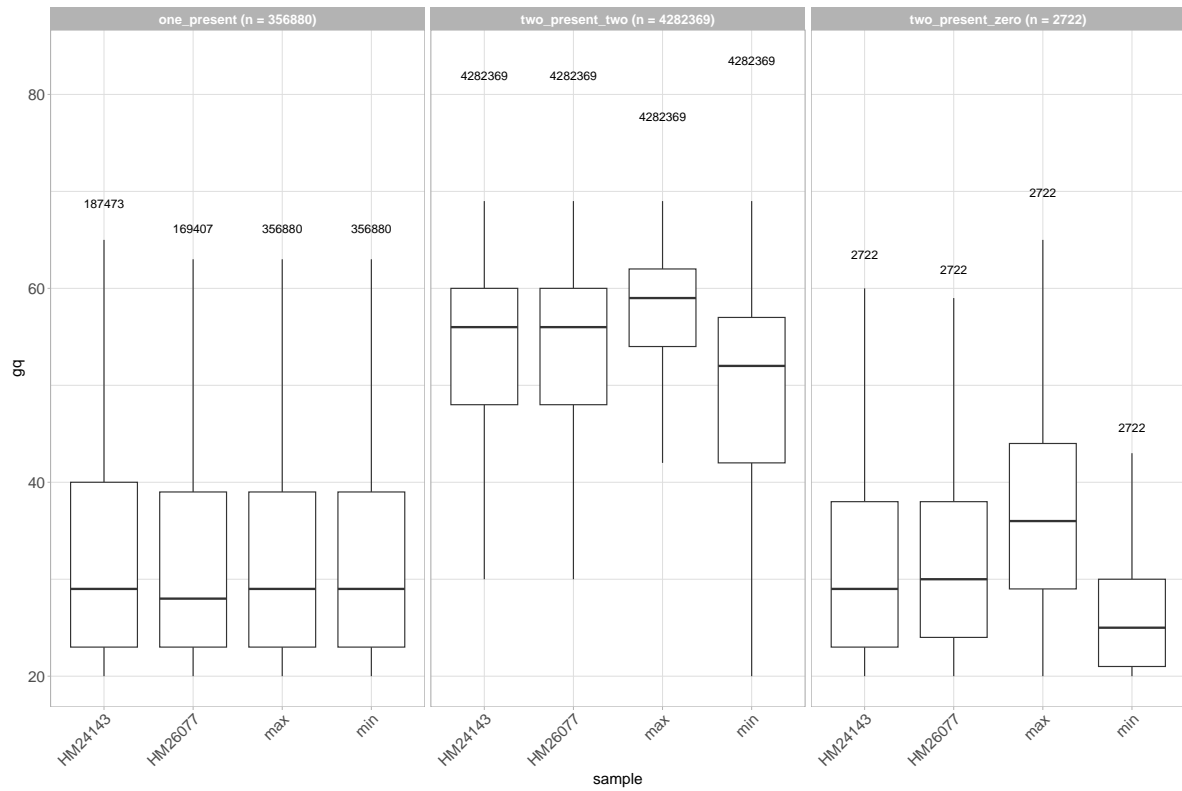
### GQ quantiles (per match status)

id	n	% total	0%	25%	50%	75%	100%
one_present	356880	7.69	20	23	29	39	69
two_present_two	4282369	92.25	20	48	56	60	69
two_present_zero	2722	0.06	20	24	30	38	67
total	4641971	—	—	—	—	—	—

### GQ quantiles (per match/sample status)

id	sample	n	% sample	0%	25%	50%	75%	100%
one_present	HM24143	187473	4.19	20	23	29	40	69
one_present	HM26077	169407	3.80	20	23	28	39	69
one_present	max	356880	7.69	20	23	29	39	69
one_present	min	356880	7.69	20	23	29	39	69
two_present_two	HM24143	4282369	95.75	20	48	56	60	69
two_present_two	HM26077	4282369	96.14	20	48	56	60	69
two_present_two	max	4282369	92.25	20	54	59	62	69
two_present_two	min	4282369	92.25	20	42	52	57	69
two_present_zero	HM24143	2722	0.06	20	23	29	38	67
two_present_zero	HM26077	2722	0.06	20	24	30	38	67
two_present_zero	max	2722	0.06	20	29	36	44	67
two_present_zero	min	2722	0.06	20	21	25	30	62

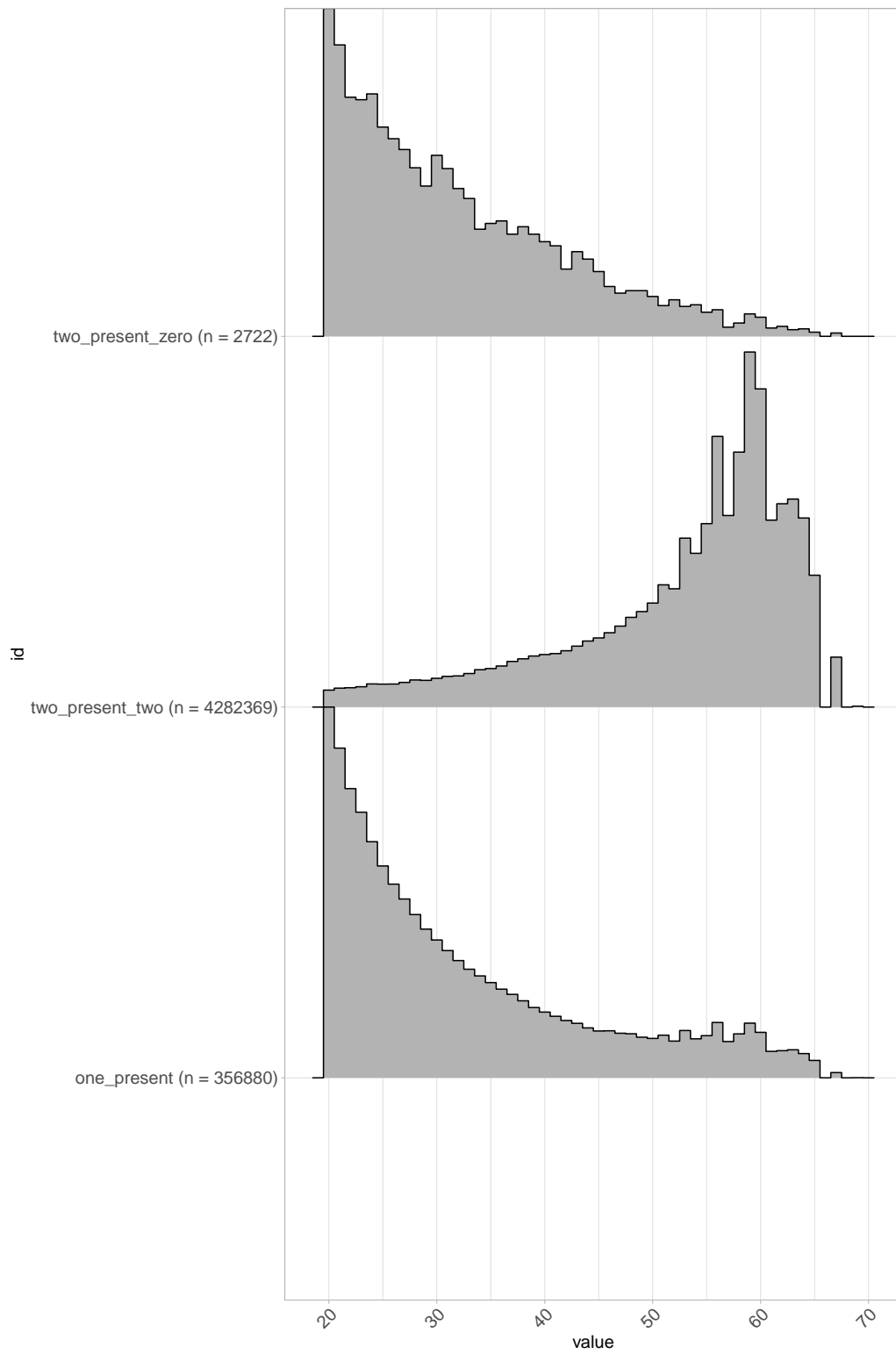
## GQ Boxplots





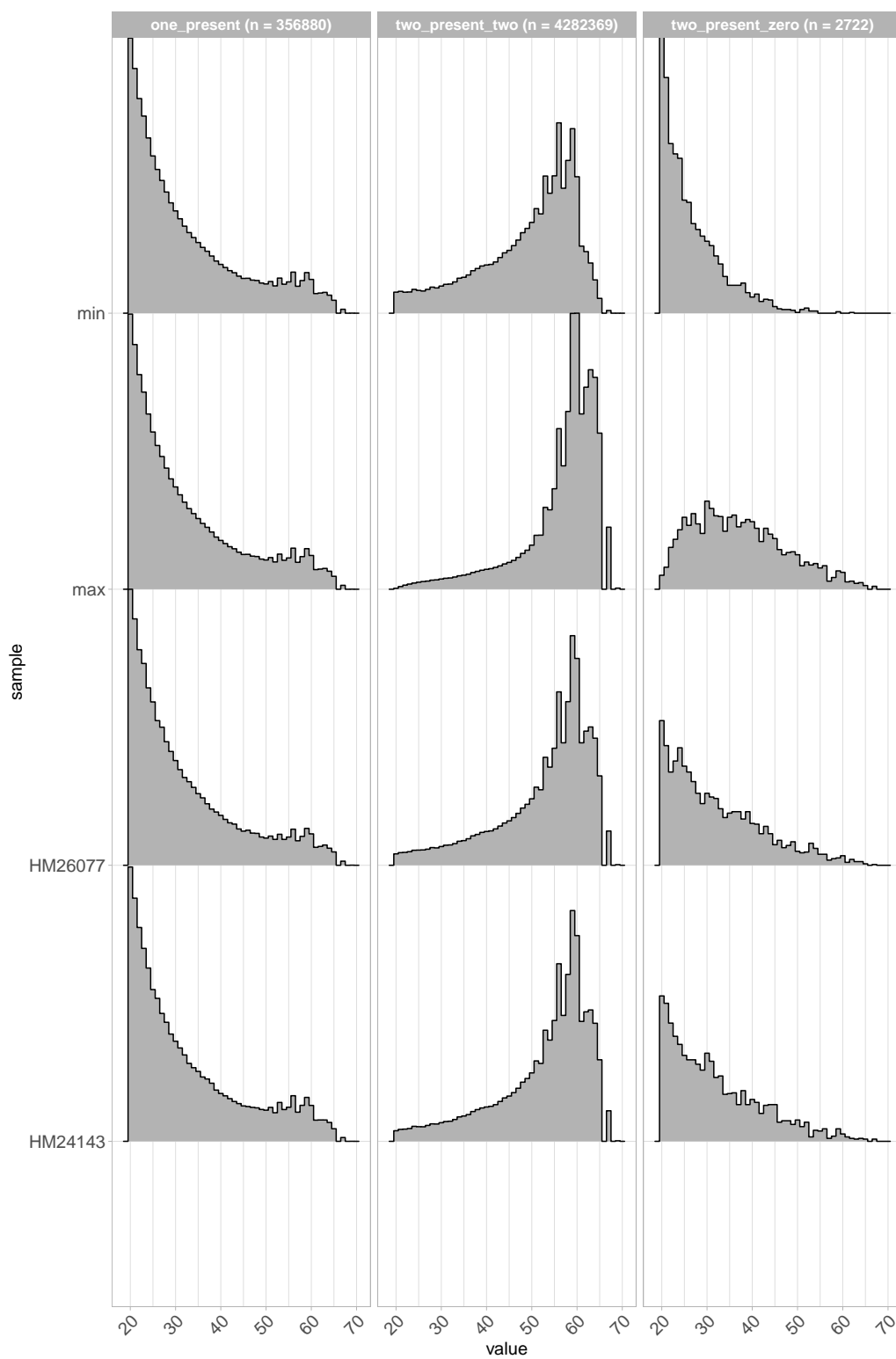


## GQ Histograms (per match status)





## GQ Histograms (per match/sample status)



## Compare HG005: HM24631, HM26107

- HM24631: LCL
- HM26107: iPSC from LCL

### Depth stats

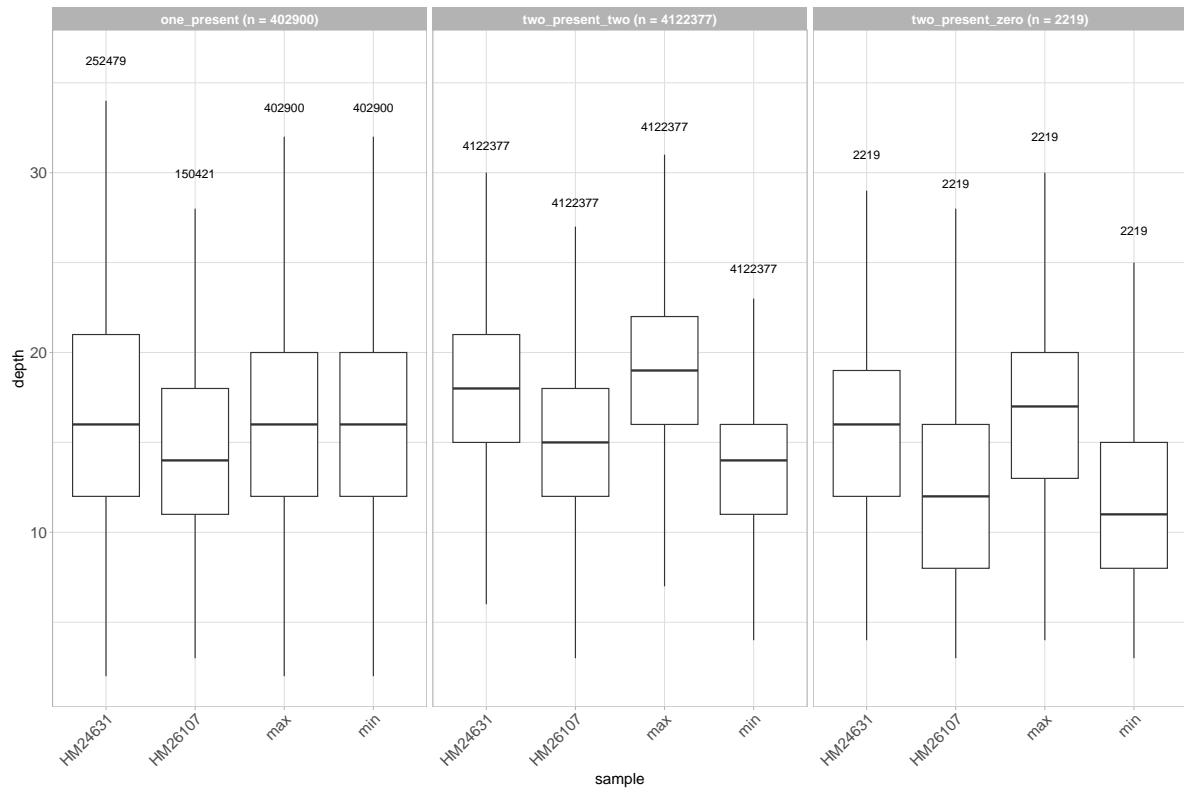
#### Depth quantiles (per match status)

id	n	% total	0%	25%	50%	75%	100%
one_present	402900	8.90	2	12	16	20	597
two_present_two	4122377	91.05	2	13	16	19	598
two_present_zero	2219	0.05	3	10	14	18	175
total	4527496	—	—	—	—	—	—

#### Depth quantiles (per match/sample status)

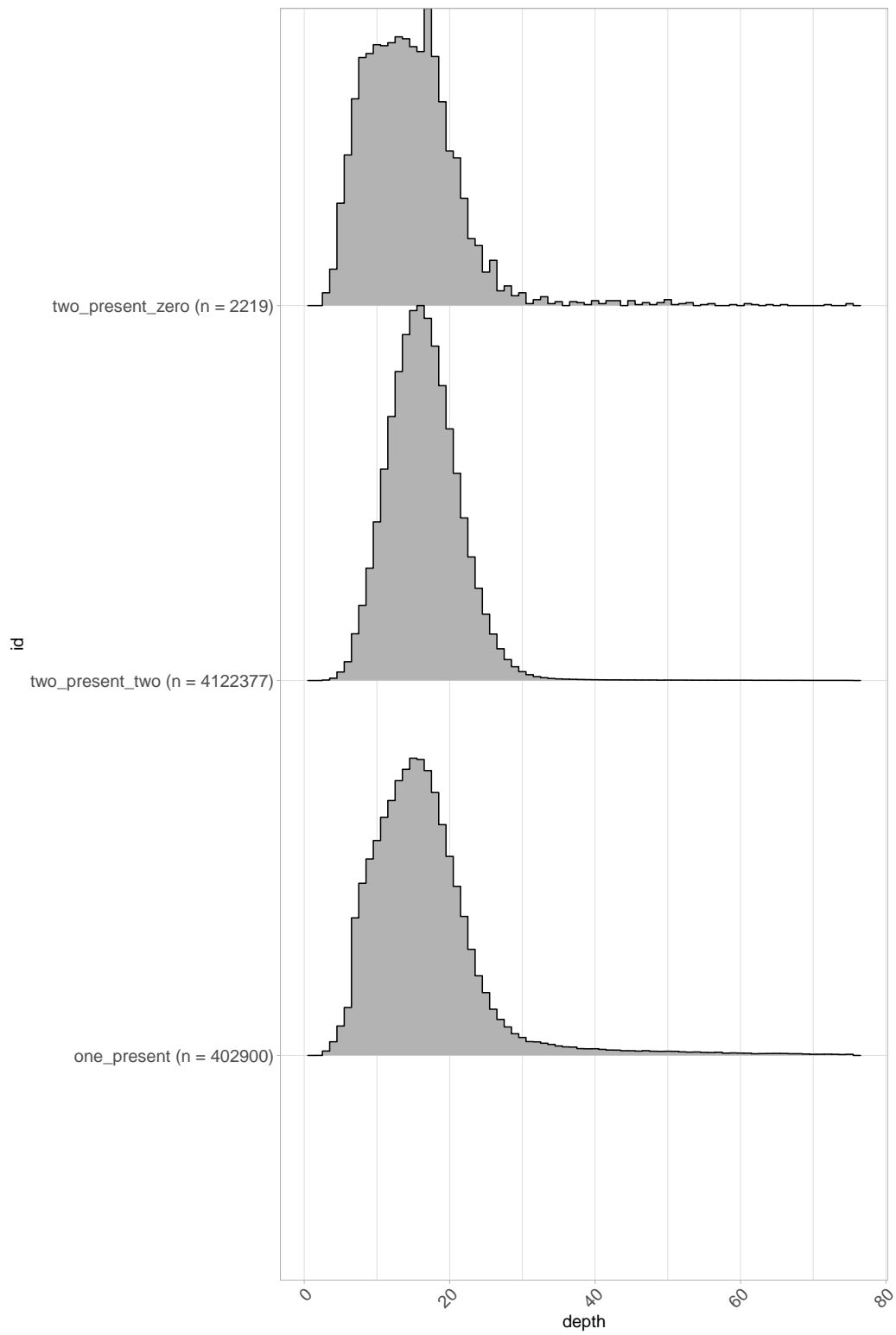
id	sample	n	% sample	0%	25%	50%	75%	100%
one_present	HM24631	252479	5.77	2	12	16	21	595
one_present	HM26107	150421	3.52	3	11	14	18	597
one_present	max	402900	8.90	2	12	16	20	597
one_present	min	402900	8.90	2	12	16	20	597
two_present_two	HM24631	4122377	94.18	2	15	18	21	597
two_present_two	HM26107	4122377	96.43	2	12	15	18	598
two_present_two	max	4122377	91.05	3	16	19	22	598
two_present_two	min	4122377	91.05	2	11	14	16	597
two_present_zero	HM24631	2219	0.05	4	12	16	19	175
two_present_zero	HM26107	2219	0.05	3	8	12	16	125
two_present_zero	max	2219	0.05	4	13	17	20	175
two_present_zero	min	2219	0.05	3	8	11	15	125

## Depth Boxplots





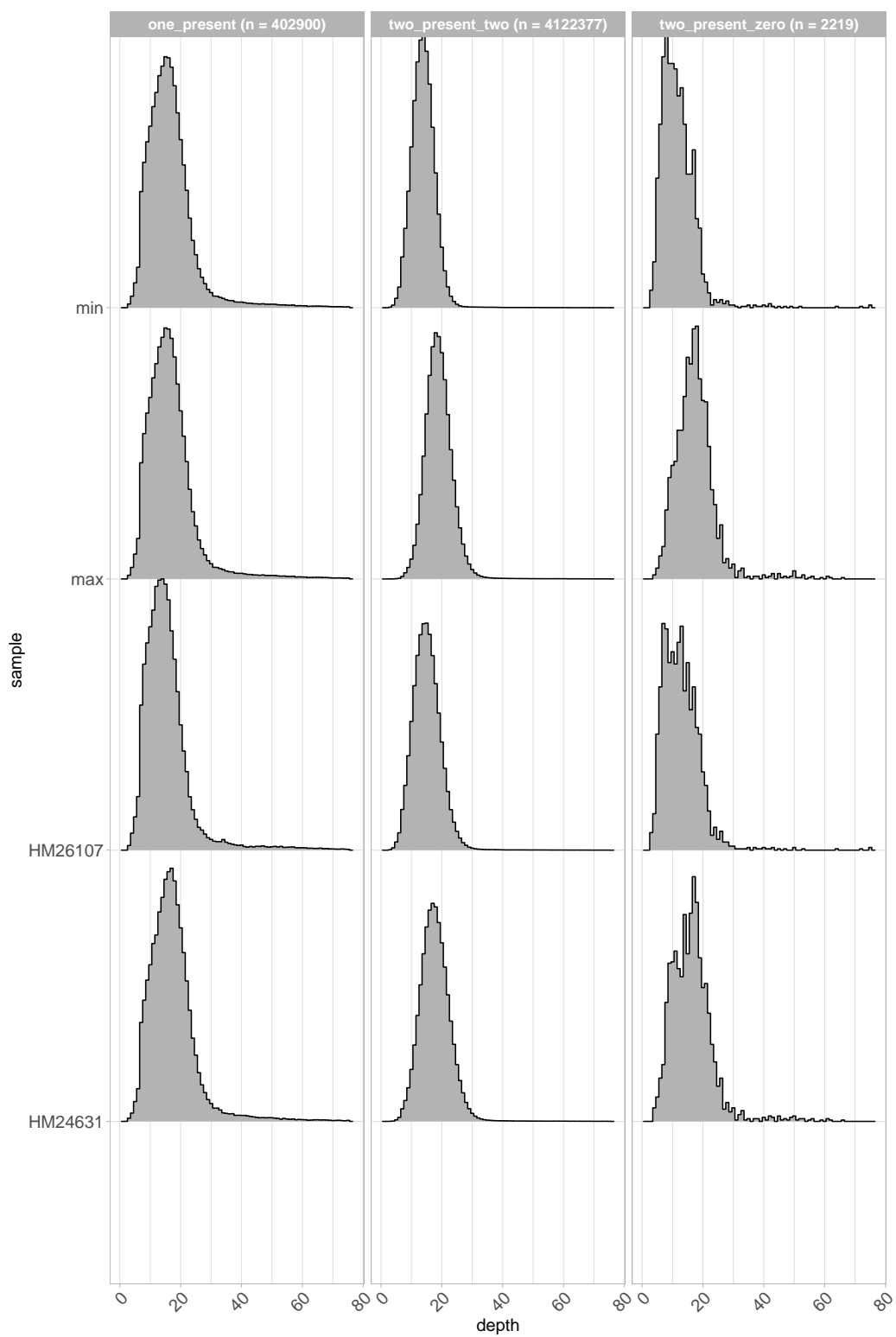
## Depth Histograms (per match status)







## Depth Histograms (per match/sample status)



## GQ stats

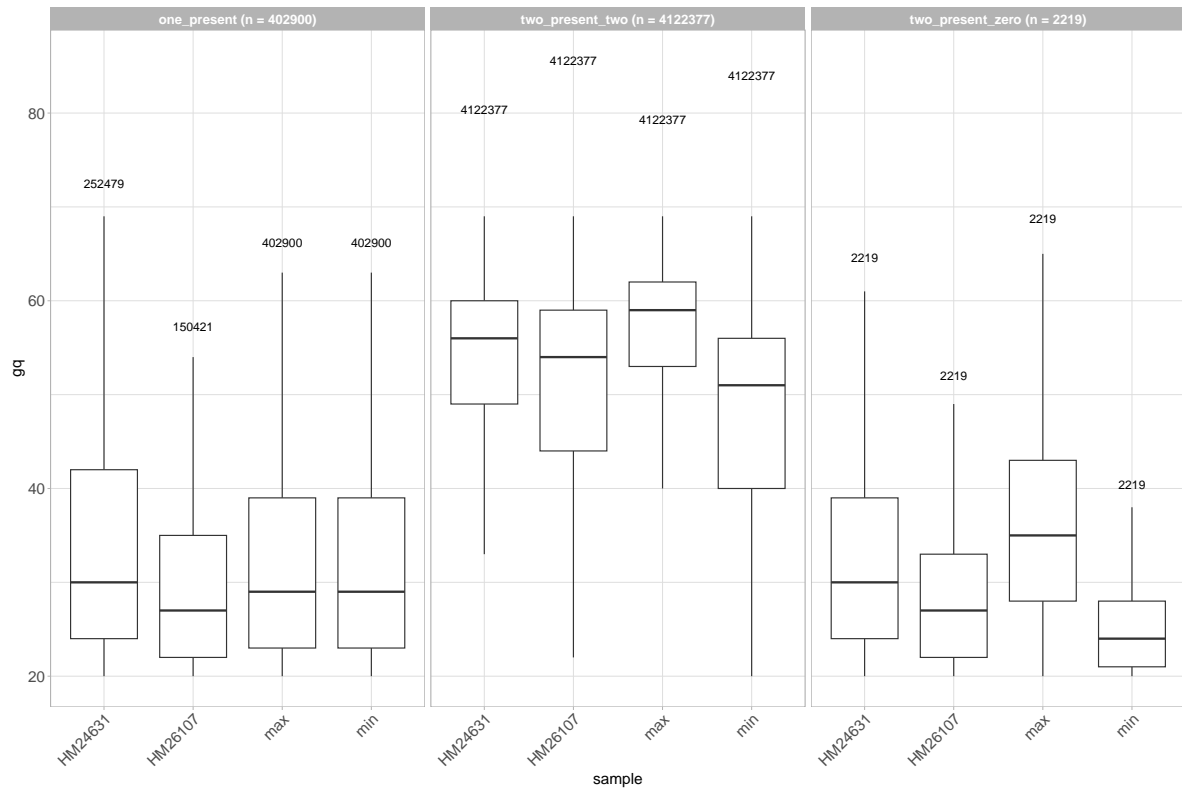
### GQ quantiles (per match status)

id	n	% total	0%	25%	50%	75%	100%
one_present	402900	8.90	20	23	29	39	69
two_present_two	4122377	91.05	20	46	55	60	69
two_present_zero	2219	0.05	20	23	28	36	69
total	4527496	—	—	—	—	—	—

### GQ quantiles (per match/sample status)

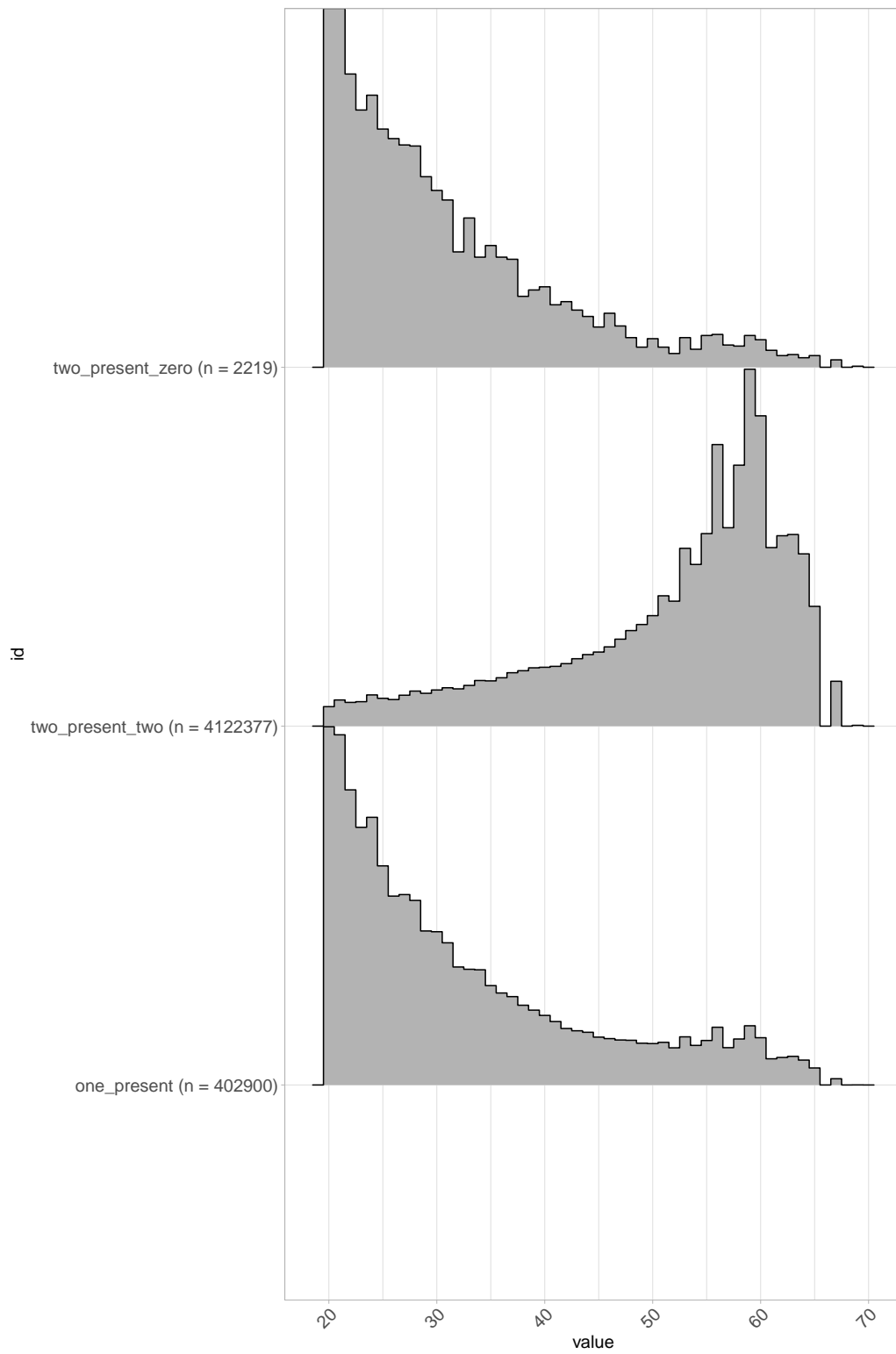
id	sample	n	% sample	0%	25%	50%	75%	100%
one_present	HM24631	252479	5.77	20	24	30	42	69
one_present	HM26107	150421	3.52	20	22	27	35	67
one_present	max	402900	8.90	20	23	29	39	69
one_present	min	402900	8.90	20	23	29	39	69
two_present_two	HM24631	4122377	94.18	20	49	56	60	69
two_present_two	HM26107	4122377	96.43	20	44	54	59	69
two_present_two	max	4122377	91.05	20	53	59	62	69
two_present_two	min	4122377	91.05	20	40	51	56	69
two_present_zero	HM24631	2219	0.05	20	24	30	39	69
two_present_zero	HM26107	2219	0.05	20	22	27	33	63
two_present_zero	max	2219	0.05	20	28	35	43	69
two_present_zero	min	2219	0.05	20	21	24	28	57

## GQ Boxplots



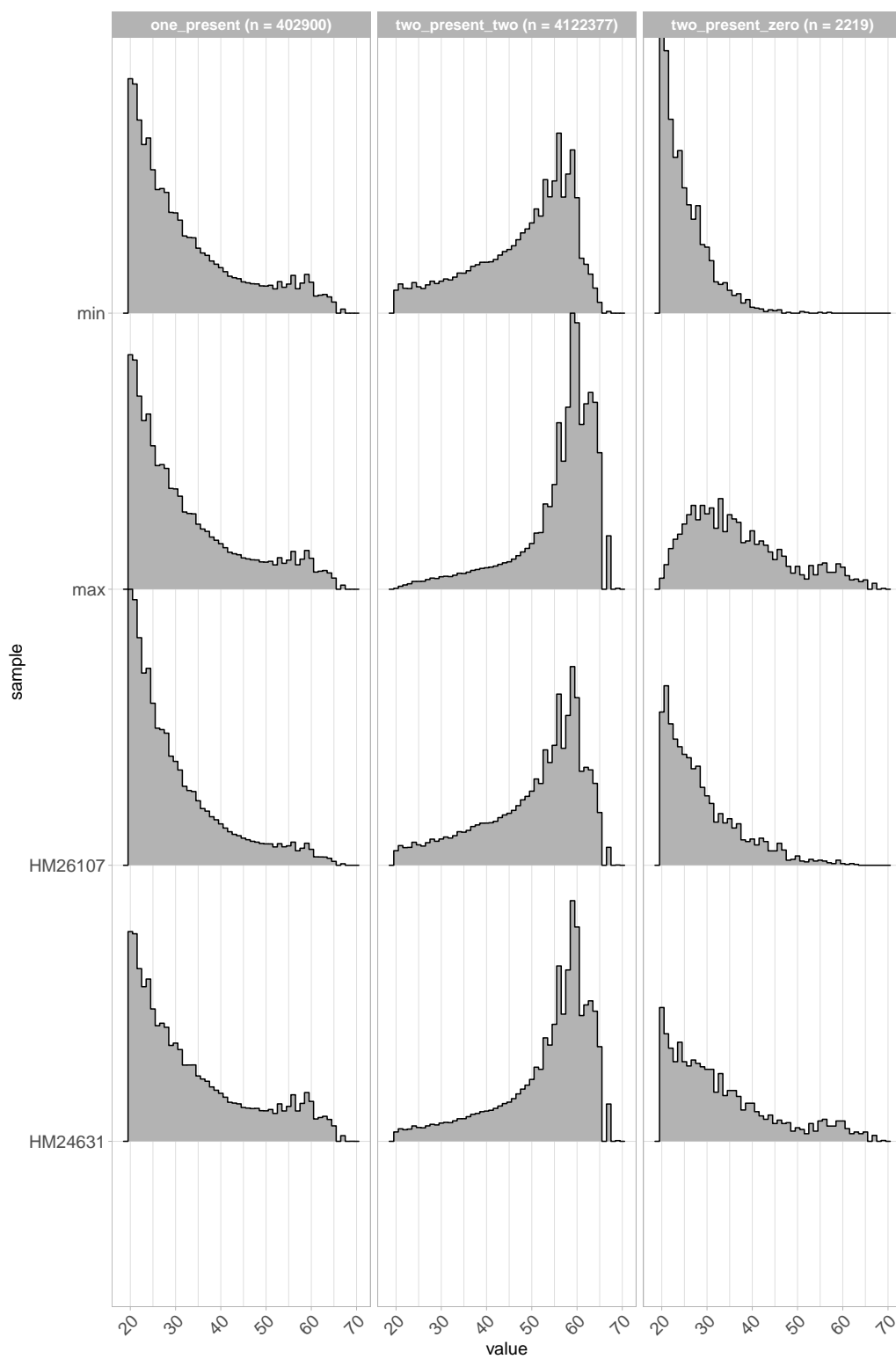


## GQ Histograms (per match status)





## GQ Histograms (per match/sample status)



## Compare Fibroblast: HM20431, HM23248\_2, HM23338

- HM20431: LCL from Fibroblast
- HM23248\_2:
- HM23338: iPSC from Fibroblast

### Depth stats

#### Depth quantiles (per match status)

id	n	% total	0%	25%	50%	75%	100%
one_present	285597	6.22	2	12.0	17	21	592
three_present_three	3440689	74.91	2	10.0	15	20	596
three_present_two	5294	0.12	2	8.0	13	19	101
three_present_zero	35	0.00	3	7.5	12	19	60
two_present_two	858229	18.68	2	13.0	17	21	586
two_present_zero	3424	0.07	3	10.0	16	20	131
total	4593268	—	—	—	—	—	—

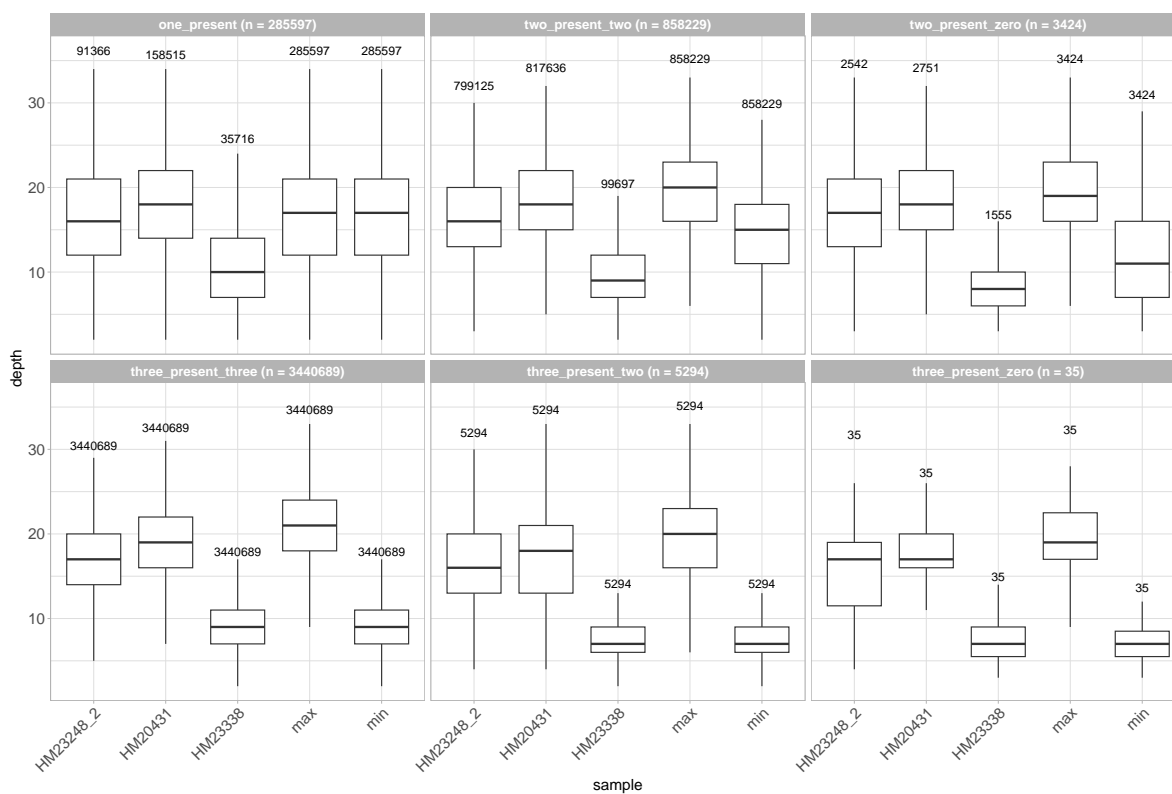
#### Depth quantiles (per match/sample status)

id	sample	n	% sample	0%	25%	50%	75%	100%
one_present	HM20431	158515	3.58	2	14.0	18	22.0	575
one_present	HM23248_2	91366	2.11	2	12.0	16	21.0	592
one_present	HM23338	35716	1.00	2	7.0	10	14.0	571
one_present	max	285597	6.22	2	12.0	17	21.0	592
one_present	min	285597	6.22	2	12.0	17	21.0	592
three_present_three	HM20431	3440689	77.76	2	16.0	19	22.0	596
three_present_three	HM23248_2	3440689	79.30	2	14.0	17	20.0	593
three_present_three	HM23338	3440689	96.03	2	7.0	9	11.0	591
three_present_three	max	3440689	74.91	4	18.0	21	24.0	596
three_present_three	min	3440689	74.91	2	7.0	9	11.0	590
three_present_two	HM20431	5294	0.12	4	13.0	18	21.0	101
three_present_two	HM23248_2	5294	0.12	4	13.0	16	20.0	101
three_present_two	HM23338	5294	0.15	2	6.0	7	9.0	47
three_present_two	max	5294	0.12	6	16.0	20	23.0	101
three_present_two	min	5294	0.12	2	6.0	7	9.0	47
three_present_zero	HM20431	35	0.00	8	16.0	17	20.0	58
three_present_zero	HM23248_2	35	0.00	4	11.5	17	19.0	60



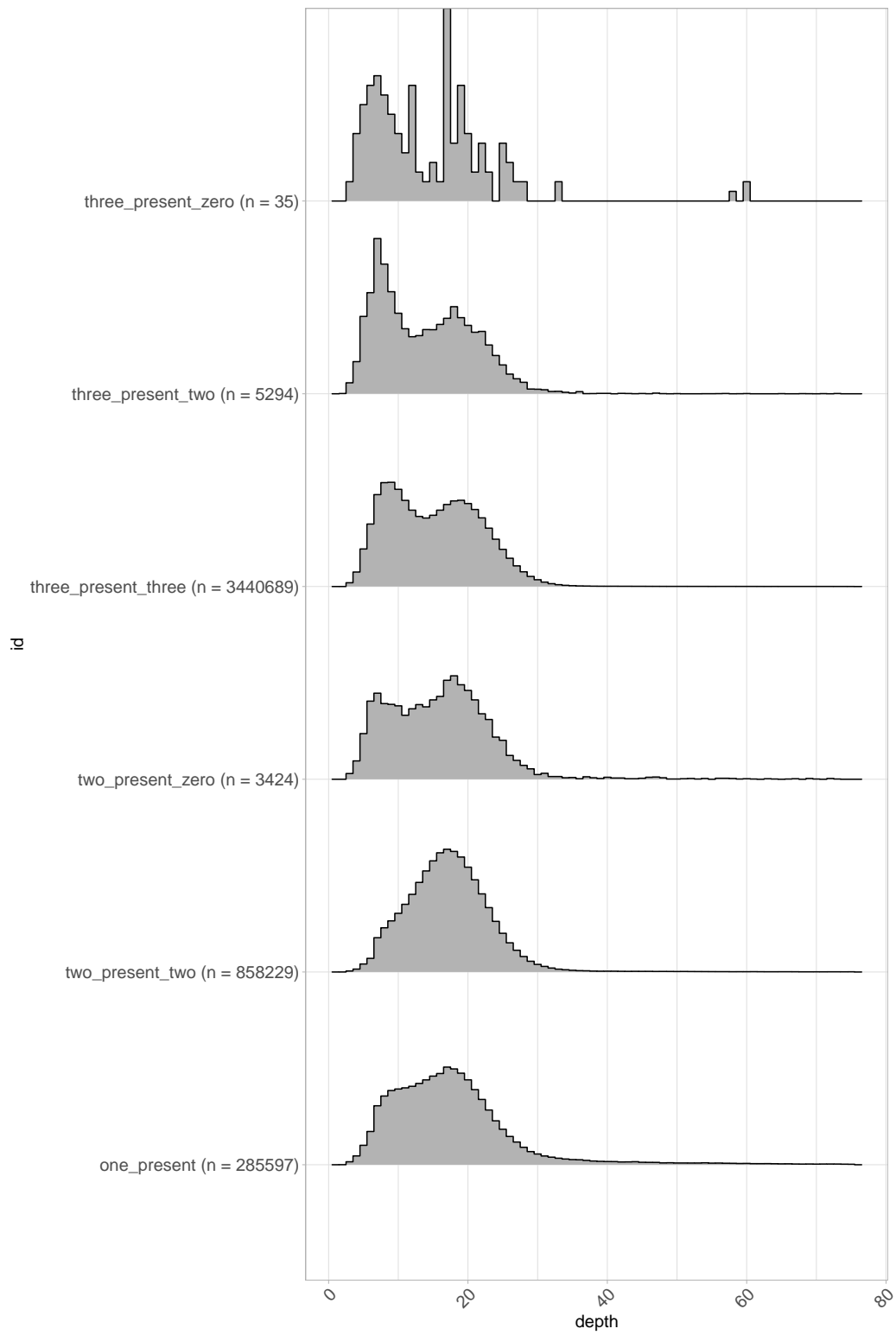
three_present_zero	HM23338	35	0.00	3	5.5	7	9.0	27
three_present_zero	max	35	0.00	9	17.0	19	22.5	60
three_present_zero	min	35	0.00	3	5.5	7	8.5	27
two_present_two	HM20431	817636	18.48	2	15.0	18	22.0	584
two_present_two	HM23248_2	799125	18.42	2	13.0	16	20.0	586
two_present_two	HM23338	99697	2.78	2	7.0	9	12.0	582
two_present_two	max	858229	18.68	3	16.0	20	23.0	586
two_present_two	min	858229	18.68	2	11.0	15	18.0	582
two_present_zero	HM20431	2751	0.06	4	15.0	18	22.0	131
two_present_zero	HM23248_2	2542	0.06	3	13.0	17	21.0	126
two_present_zero	HM23338	1555	0.04	3	6.0	8	10.0	48
two_present_zero	max	3424	0.07	4	16.0	19	23.0	131
two_present_zero	min	3424	0.07	3	7.0	11	16.0	126

## Depth Boxplots



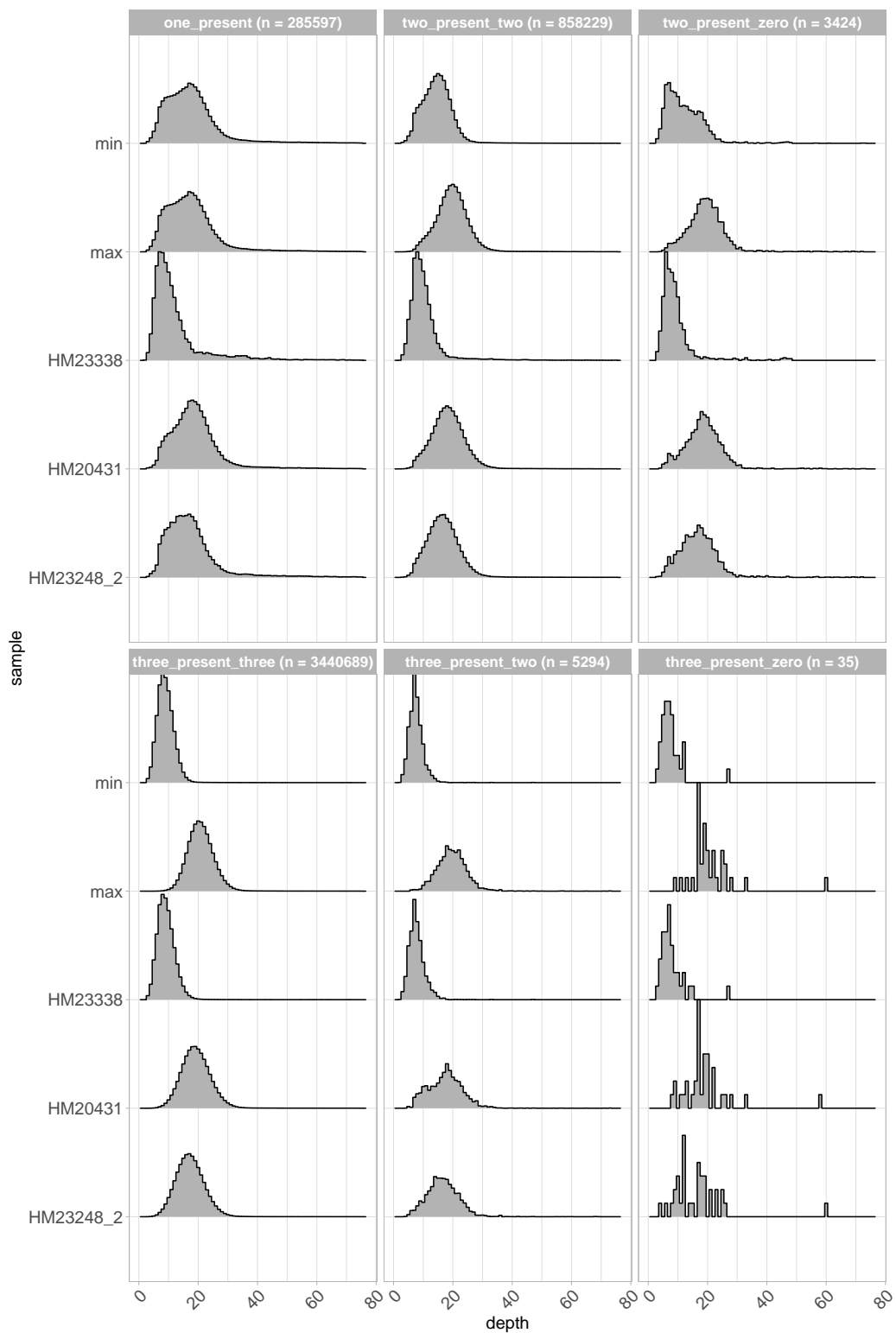


## Depth Histograms (per match status)





## Depth Histograms (per match/sample status)



## GQ stats

### GQ quantiles (per match status)

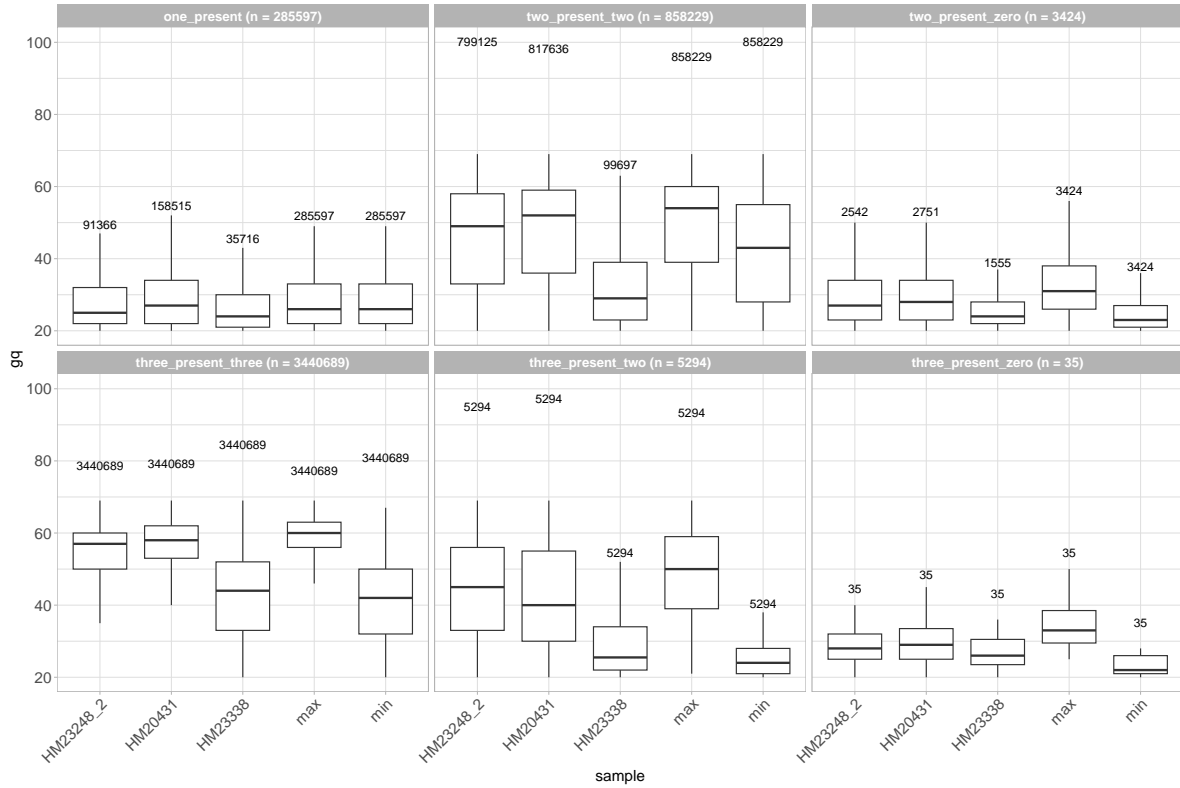
id	n	% total	0%	25%	50%	75%	100%
one_present	285597	6.22	20	22	26	33	67
three_present_three	3440689	74.91	20	43	54	60	69
three_present_two	5294	0.12	20	25	34	51	69
three_present_zero	35	0.00	20	24	28	33	50
two_present_two	858229	18.68	20	33	49	58	69
two_present_zero	3424	0.07	20	22	26	33	67
total	4593268	—	—	—	—	—	—

### GQ quantiles (per match/sample status)

id	sample	n	% sample	0%	25%	50%	75%	100%
one_present	HM20431	158515	3.58	20	22.0	27.0	34.0	67
one_present	HM23248_2	91366	2.11	20	22.0	25.0	32.0	67
one_present	HM23338	35716	1.00	20	21.0	24.0	30.0	64
one_present	max	285597	6.22	20	22.0	26.0	33.0	67
one_present	min	285597	6.22	20	22.0	26.0	33.0	67
three_present_three	HM20431	3440689	77.76	20	53.0	58.0	62.0	69
three_present_three	HM23248_2	3440689	79.30	20	50.0	57.0	60.0	69
three_present_three	HM23338	3440689	96.03	20	33.0	44.0	52.0	69
three_present_three	max	3440689	74.91	20	56.0	60.0	63.0	69
three_present_three	min	3440689	74.91	20	32.0	42.0	50.0	67
three_present_two	HM20431	5294	0.12	20	30.0	40.0	55.0	69
three_present_two	HM23248_2	5294	0.12	20	33.0	45.0	56.0	69
three_present_two	HM23338	5294	0.15	20	22.0	25.5	34.0	65
three_present_two	max	5294	0.12	21	39.0	50.0	59.0	69
three_present_two	min	5294	0.12	20	21.0	24.0	28.0	58
three_present_zero	HM20431	35	0.00	20	25.0	29.0	33.5	50
three_present_zero	HM23248_2	35	0.00	20	25.0	28.0	32.0	45
three_present_zero	HM23338	35	0.00	20	23.5	26.0	30.5	46
three_present_zero	max	35	0.00	25	29.5	33.0	38.5	50
three_present_zero	min	35	0.00	20	21.0	22.0	26.0	36
two_present_two	HM20431	817636	18.48	20	36.0	52.0	59.0	69
two_present_two	HM23248_2	799125	18.42	20	33.0	49.0	58.0	69
two_present_two	HM23338	99697	2.78	20	23.0	29.0	39.0	69
two_present_two	max	858229	18.68	20	39.0	54.0	60.0	69

two_present_two	min	858229	18.68	20	28.0	43.0	55.0	69
two_present_zero	HM20431	2751	0.06	20	23.0	28.0	34.0	65
two_present_zero	HM23248_2	2542	0.06	20	23.0	27.0	34.0	67
two_present_zero	HM23338	1555	0.04	20	22.0	24.0	28.0	60
two_present_zero	max	3424	0.07	20	26.0	31.0	38.0	67
two_present_zero	min	3424	0.07	20	21.0	23.0	27.0	58

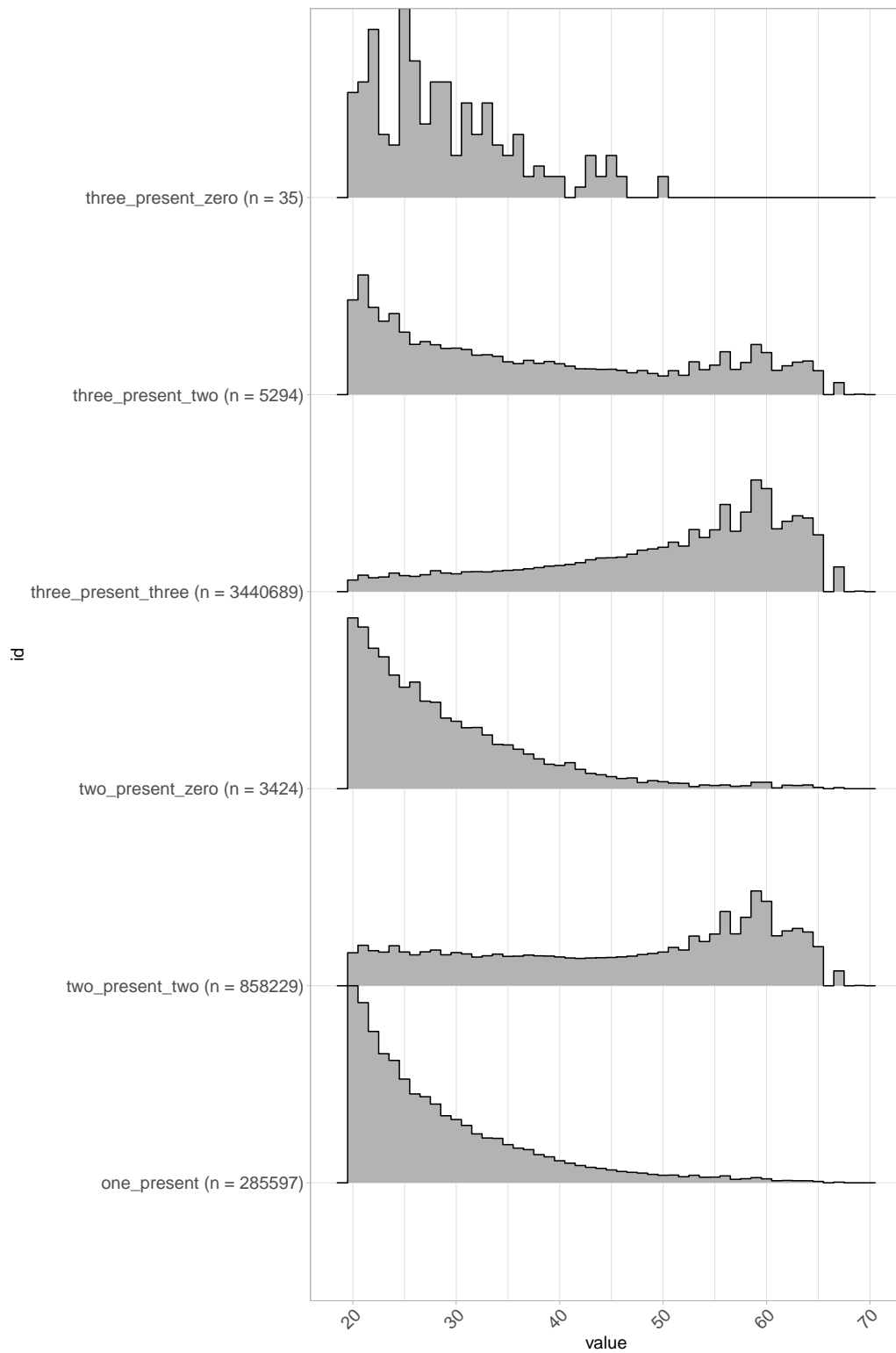
## GQ Boxplots





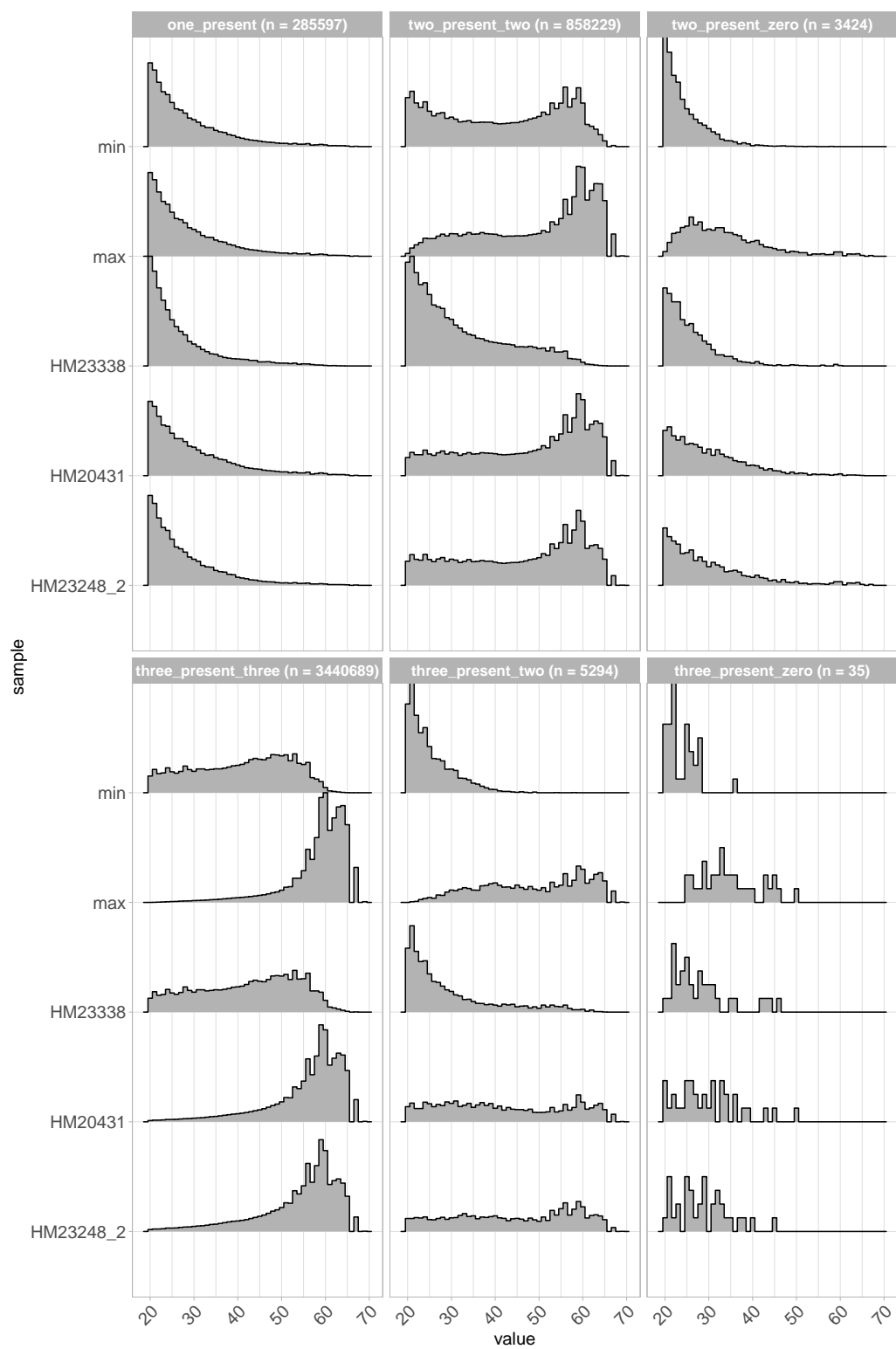


## GQ Histograms (per match status)





## GQ Histograms (per match/sample status)





## CYP2D6 Coverage

