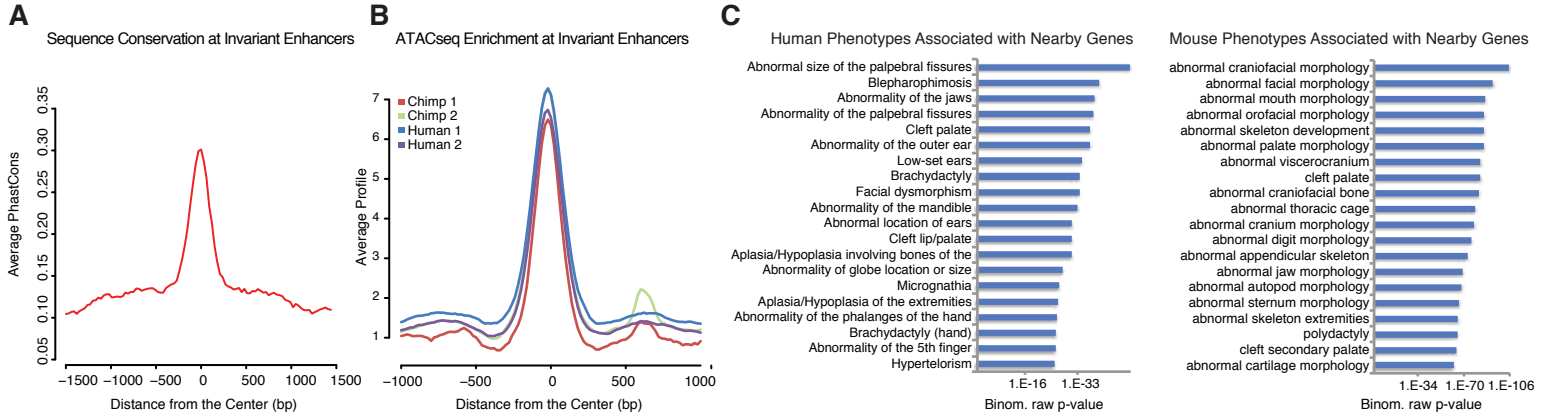
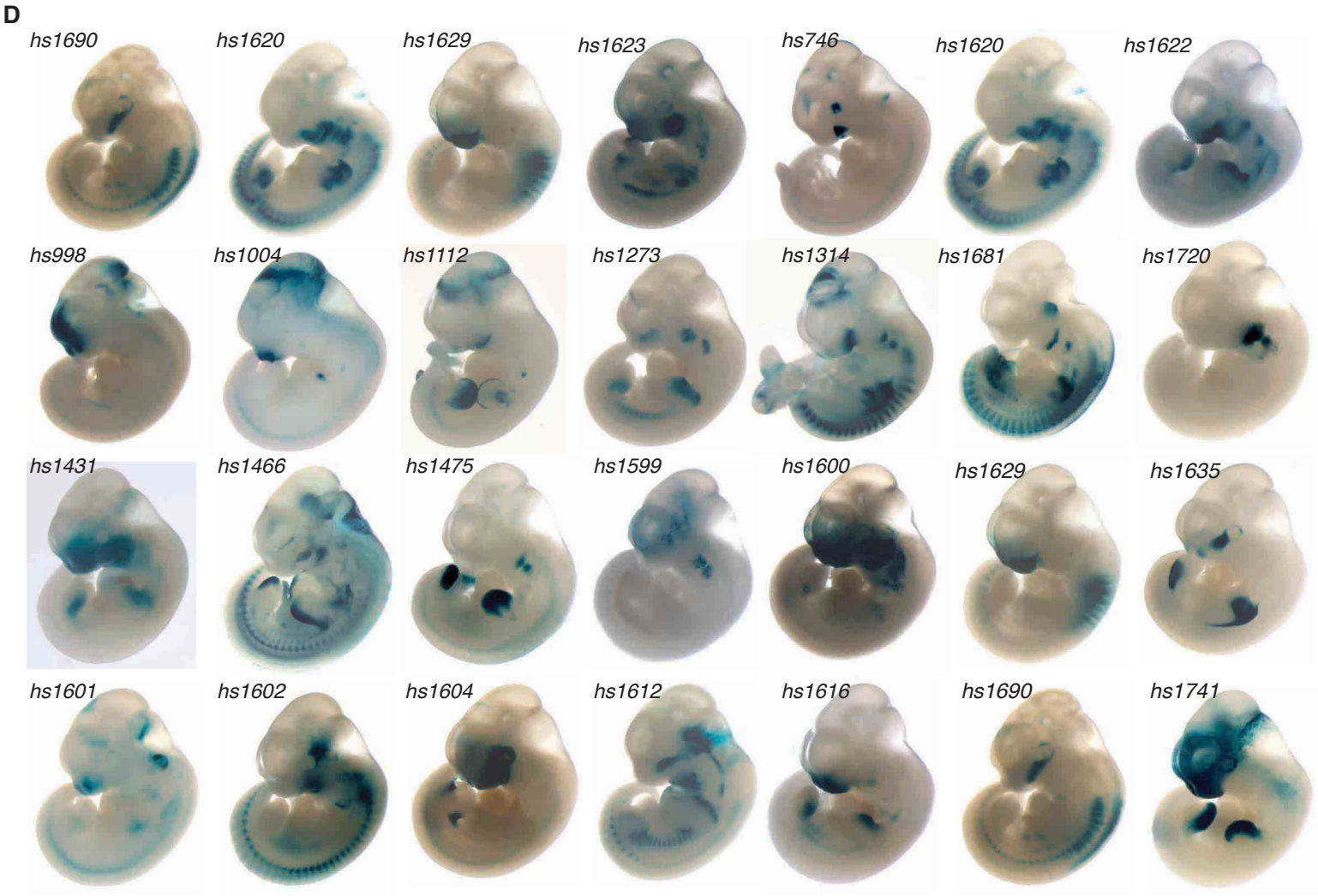

A
Sequence Conservation at Invariant Enhancers
Average PhastCons
0.05
0.10
0.15
0.20
0.25
0.30
0.35
−1500 −1000 −500 0 500 1000 1500
Distance from the Center (bp)
B
ATACseq Enrichment at Invariant Enhancers
Chimp 1
Chimp 2
Human 1
Human 2
Average Profile
1
2
3
4
5
6
7
−1000 −500 0 500 1000
Distance from the Center (bp)
C
Human Phenotypes Associated with Nearby Genes
Abnormal size of the palpebral fissures
Blepharophimosis
Abnormality of the jaws
Abnormality of the palpebral fissures
Cleft palate
Abnormality of the outer ear
Low-set ears
Brachydactyly
Facial dysmorphism
Abnormality of the mandible
Abnormal location of ears
Cleft lip/palate
Aplasia/Hypoplasia involving bones of the
Abnormality of globe location or size
Micrognathia
Aplasia/Hypoplasia of the extremities
Abnormality of the phalanges of the hand
Brachydactyly (hand)
Abnormality of the 5th finger
Hypertelorism
1.E-16 1.E-33
Binom. raw p-value
Mouse Phenotypes Associated with Nearby Genes
abnormal craniofacial morphology
abnormal facial morphology
abnormal mouth morphology
abnormal orofacial morphology
abnormal skeleton development
abnormal palate morphology
abnormal viscerocranium
cleft palate
abnormal craniofacial bone
abnormal thoracic cage
abnormal cranium morphology
abnormal digit morphology
abnormal appendicular skeleton
abnormal jaw morphology
abnormal autopod morphology
abnormal sternum morphology
abnormal skeleton extremities
polydactyly
cleft secondary palate
abnormal cartilage morphology
1.E-34 1.E-70 1.E-106
Binom. raw p-value


D
hs1690
hs1620
hs1629
hs1623
hs746
hs1620
hs1622
hs998
hs1004
hs1112
hs1273
hs1314
hs1681
hs1720
hs1431
hs1466
hs1475
hs1599
hs1600
hs1629
hs1635
hs1601
hs1602
hs1604
hs1612
hs1616
hs1690
hs1741