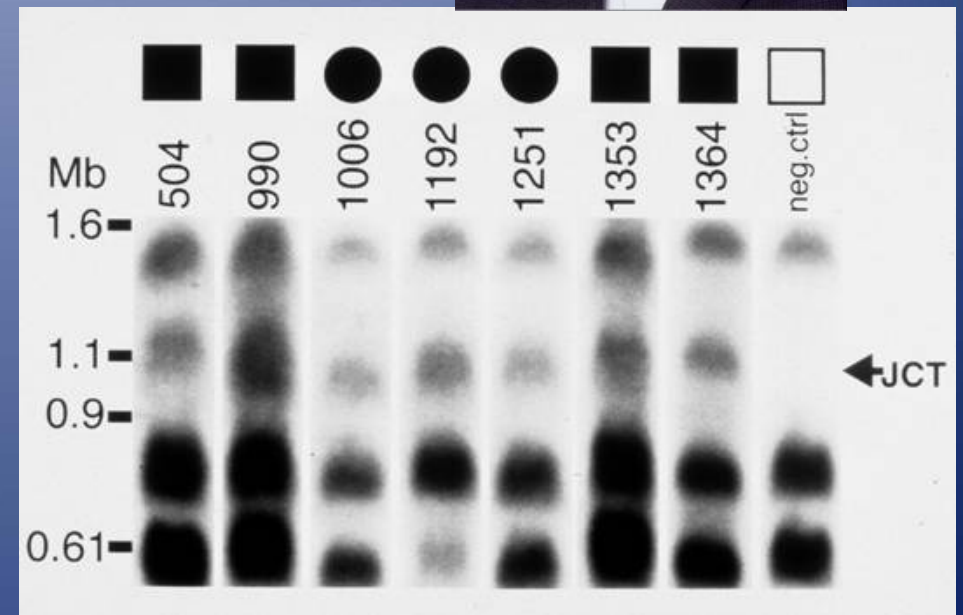
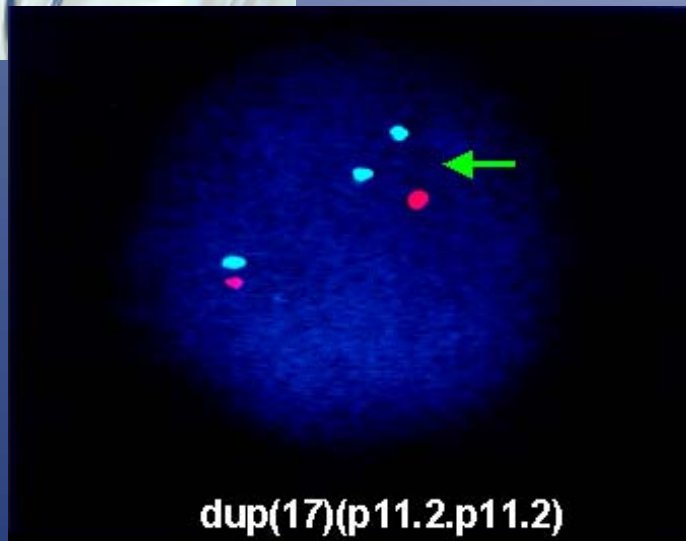
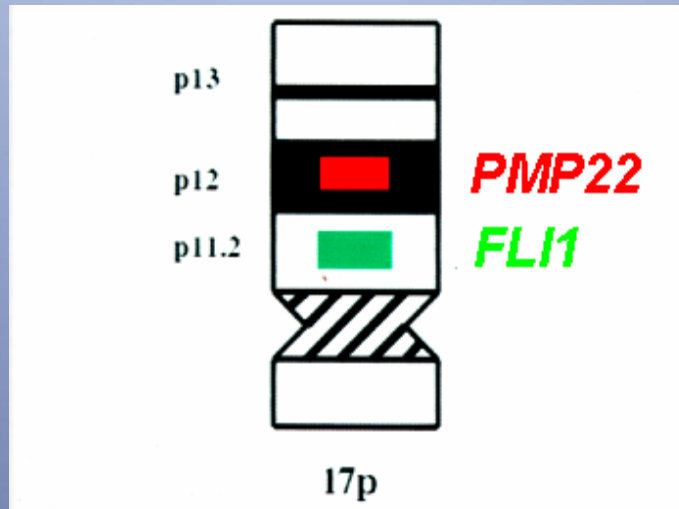


Potocki-Lupski syndrome (PTLS;MIM #610883)



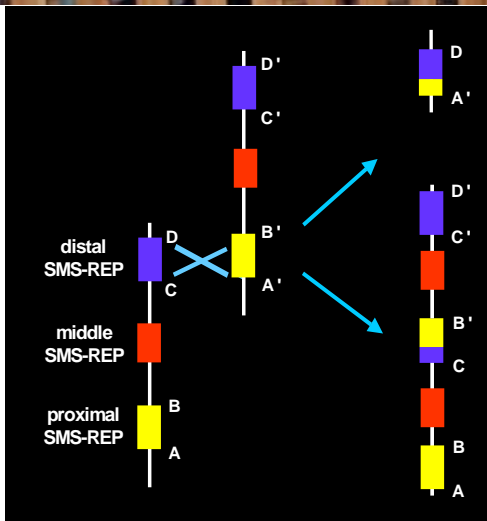
2000, seven patients with common duplication were described;
2007, multidisciplinary clinical study

Potocki et al (2000) *Nat Genet* 24:84-87
Potocki et al (2007) *AJHG* 80:633-649



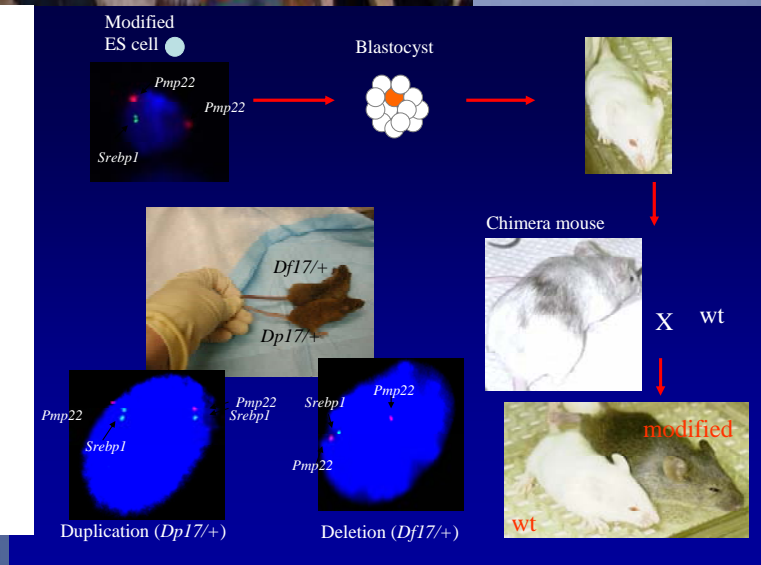
Potocki-Lupski Syndrome

(PTLS;
OMIM
#610883)

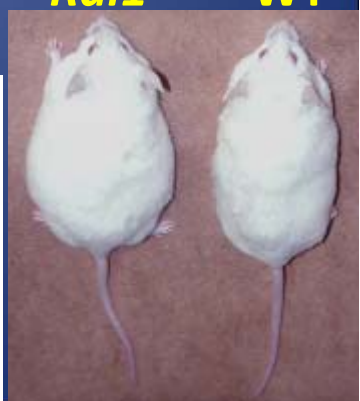
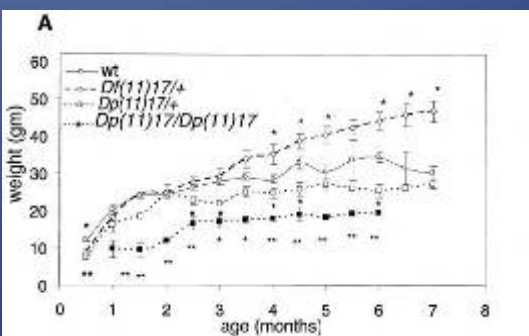
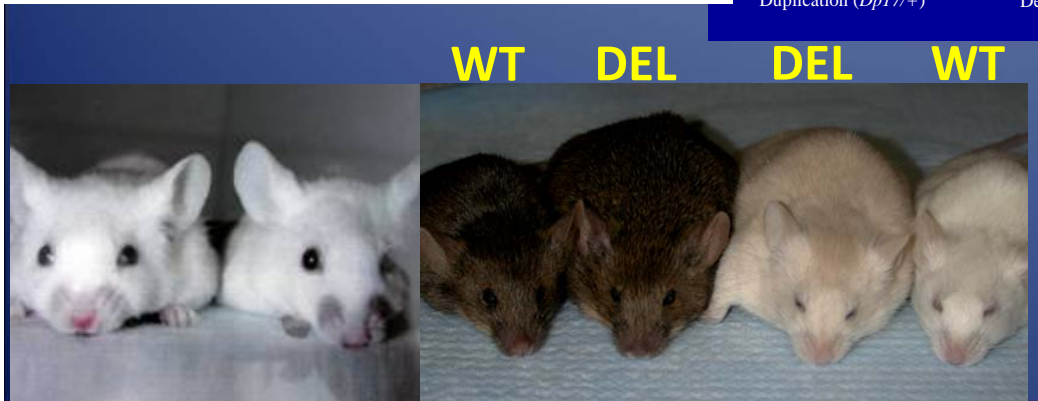


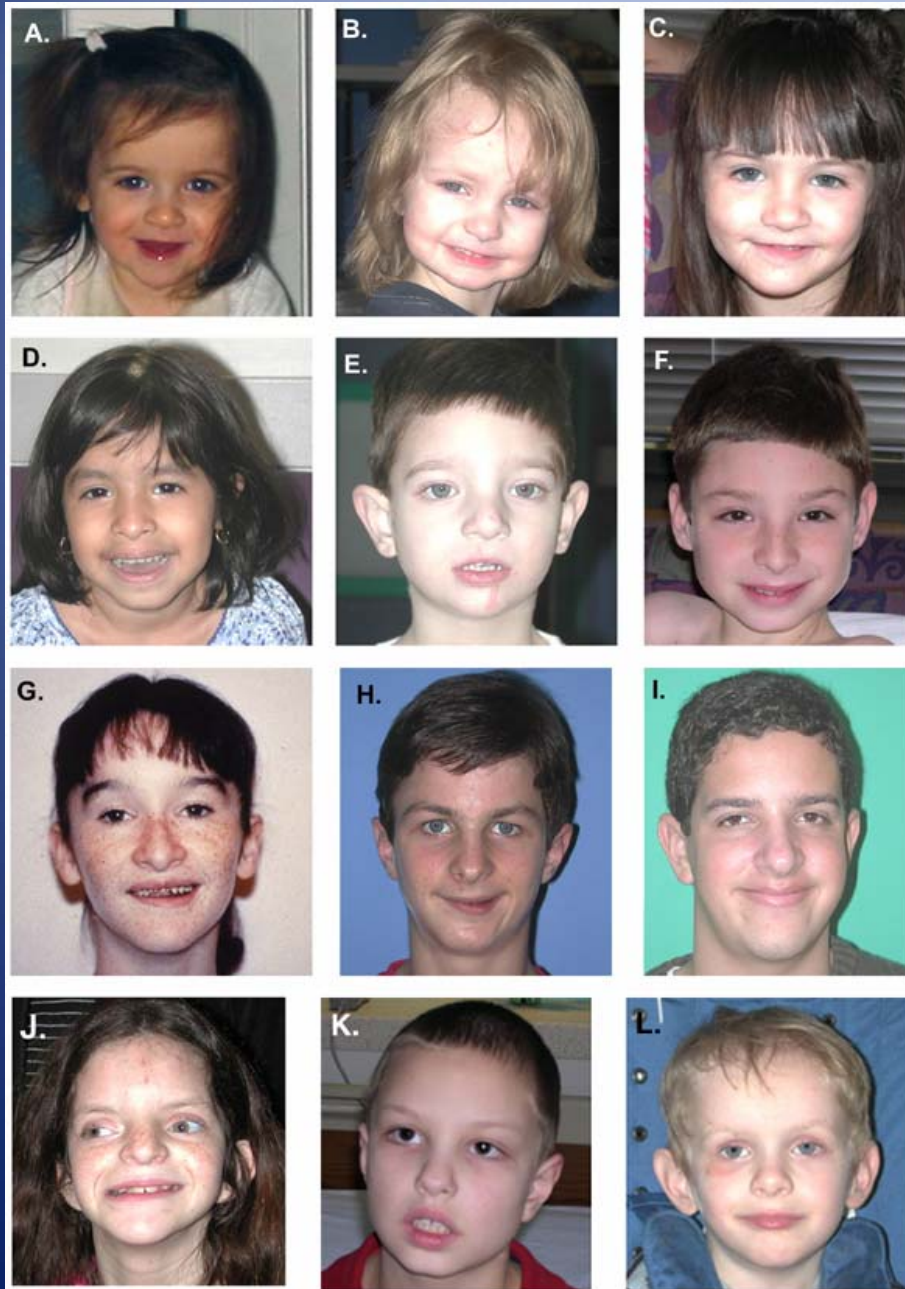
SMS
common
deletion

PTLS
common
duplication



Rai1^{+/-} WT





Potocki-Lupski syndrome (PTLS;MIM #610883)

dup(17)(p11.2p11.2)

- Less severe than del17p11.2
- Major features
 - MR/DD, hypotonia
 - Craniofacial features
 - Triangular face
 - Neurobehavioral abnormalities
 - Hyperactivity
 - Attention deficit
 - **Autistic features, RAI1 ***
 - Dental abnormalities

Potocki et al. (2007) *AJHG* 80:633-649

Treadwell-Deering *et al.* (2010) *J Dev Behav Pediatr.* 31 :137-43.

*** RAI1 highest prioritizer analysis**
P-value [0.0012] of non-complex autism
patient CNVs; van der Zwaag, *et al* (2009)
***PLoS ONE* 4 :e5324**