Table 2. Notch signaling and developmental diseases

Notch Component	Inheritance	Molecular Mechanism	Disease
Delta-like-3 (DLL-3) 19q13.2	autosomal recessive	Mutations on DLL-3 usually lead to expression of a truncated protein or to amino acid substitutions	Spondylocostal dysostosis type1 (trunk dwarfism secondary to rib anomalies and vertebral segmentation defects)
Mesoderm posterior 2 (MESP2) 15q26.1	autosomal recessive	Mutation in MESP2 produces a non-functional protein susceptible to nonsense-mediated RNA decay, up-regulating Notch signaling	Spondylocostal dysostosis type 2 (segmentation abnormalities of the thoracic vertebrae)
Lunatic Fringe (LFNG) 7p22	autosomal recessive	Missense mutations of LFNG up-regulates Notch signaling	Spondylocostal dysostosis type 3
Hairy Enhancer of Split 7 (HES7) 17p13.1	autosomal recessive	Missense mutations of HES7 up-regulates Notch signaling	Spondylocostal dysostosis type 4
Chondroitin sulfate synthase (CHSY)1*	autosomal recessive	Loss-of-function mutations lead to up-regulation of JAG-1	Recessive brachydactyly
JAG-1 (20p12.2), NOTCH 2 (1p12)	autosomal dominant	Heterozygous mutations	Alagille syndrome type 1 and type 2

*Encodes a transmembrane protein which contains a Fringe domain.