DeLange syndrome

Dutch pediatrician *Cornelia Catharina De Lange* described two syndromes named De Lange Syndrome I and II (CDLS 1 and 2).[1]

Genetics

- Most cases are sporadic.
- In addition, the syndrome is genetically heterogeneous[2].
- **CDLS 1** is caused by a mutation in geneNIPBL (5p13.1) and shows autosomal dominant inheritance[2].
- **CDLS 2** is caused by a mutation in the SMC1A gene (Xp11.22-p11.21) and shows X-linked inheritance[2].

CDLS 1

Characteristics:

- Diffuse muscle hypertrophy;
- extrapyramidal disorders;
- psychomotor retardation.

CDLS 2

Characteristics:

- Brachycephaly;
- thick fused eyebrows;
- long eyelashes;
- epicants;
- hypoplastic lower jaw;
- oligophrenia;
- spina bifida occulta;
- syndactyly.

Links

References

1. ENERSEN, Ole Daniel. *Whonamed - Cornelia Catharina de Lange* [online]. [cit. 2011-05-03]. <http://www.whonamed.com/doctor.cfm/1059.html>.
2. *OMIM : CORNELIA DE LANGE SYNDROME* [database]. Johns Hopkins University. [cit. 2011-05-03]. <http://omim.org/entry/122470>.

References

- LAZOVSKIS, Ilmars – DOBIÁŠ, Wenceslas. *Overview of clinical symptoms and syndromes.* 2. edition. Avicenum, healthcare publishing house, 1990. pp. 581. ISBN 80-201-0043-1.