



HAL
open science

Asphyxiating Thoracic Dysplasia: clinical and molecular review of 42 families

Valérie Cormier-Daire, Céline Huber, Geneviève Baujat, R Caumes, Honorine Kayirangwa, Martine Le Merrer, Kim-Hanh Le Quan Sang, Arnold Munnich

► **To cite this version:**

Valérie Cormier-Daire, Céline Huber, Geneviève Baujat, R Caumes, Honorine Kayirangwa, et al.. Asphyxiating Thoracic Dysplasia: clinical and molecular review of 42 families. First International Cilia in Development and Disease Scientific Conference, pp.O4. inserm-00752961

HAL Id: inserm-00752961

<https://inserm.hal.science/inserm-00752961>

Submitted on 16 Nov 2012

HAL is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.

ORAL PRESENTATION

Open Access

Asphyxiating Thoracic Dysplasia: clinical and molecular review of 42 families

V Cormier-Daire*, C Huber, J Baujat, R Caumes, H Kayirangwa, M Le Merrer, KH Le Quan Sang, A Munnich

From First International Cilia in Development and Disease Scientific Conference (2012)
London, UK. 16-18 May 2012

Asphyxiating Thoracic Dysplasia (ATD) ((MIM 208500, MIM 6112633, MIM 613091, MIM 61819, MIM614376) belongs to the short rib polydactyly group and is characterized by a long and narrow thorax, short long bones and trident acetabular roof. Polydactyly, retinal degeneration, cystic renal and liver diseases have been occasionally reported. Today, mutations in *IFT80* (MIM 611177), *DYNC2H1* (MIM 603297), *TCC21B* (MIM 612014) and *WDR19* (MIM 608151) genes have been reported in ATD. Through a national grant (PHRC, AOM 06031), we have collected 55 ATD cases including 29 fetuses issued from 42 families who benefit the combined approach of deep phenotyping and molecular screening of *IFT80* and *DYNC2H1*. The series included 26 alive cases ranging in age from 6 months to 36 years. Respiratory treatment was needed in 46%, including positive pressure respiration, and invasive or non-invasive ventilation. Cystic renal and liver diseases occur in 16% of cases; whereas retinal degeneration was present in 40 % cases aged more than 2 years (6/15). The molecular screening allowed us to detect *DYNC2H1* mutations in 63% and *IFT80* mutations in 6%. In 6 cases, only one heterozygote mutation in either *IFT80* or *DYNC2H1* was identified. Finally, the two genes were excluded in 31% cases. These preliminary results emphasize that *DYNC2H1* is the major gene responsible for ATD. The presence of only one mutation (27% of mutated cases) may suggest a digenic diallelic inheritance. Ongoing studies will hopefully lead to the identification of other disease genes.

Published: 16 November 2012

* Correspondence: valerie.cormier-daire@inserm.fr
Département de Génétique, INSERM U781, Hôpital Necker Enfants Malades, France

doi:10.1186/2046-2530-1-S1-O4

Cite this article as: Cormier-Daire et al: Asphyxiating Thoracic Dysplasia: clinical and molecular review of 42 families. *Cilia* 2012 1(Suppl 1):O4.

Submit your next manuscript to BioMed Central and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar
- Research which is freely available for redistribution

Submit your manuscript at
www.biomedcentral.com/submit

