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## Pathologic expansion in the *C9orf72* gene is associated with accelerated decline of respiratory function and decreased survival in amyotrophic lateral sclerosis

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### ABSTRACT

**Introduction:** Respiratory insufficiency is the main cause of death in amyotrophic lateral sclerosis (ALS). As the *C9orf72* repeat expansion represents the most common genetic risk factor for this disease, we studied whether *C9orf72* modulates respiratory function and survival.

**Methods:** Demographic and clinical data, and *C9orf72* status were collected from 372 ALS patients followed in our centre. Multiple regressions controlling for the *C9orf72* expansion, diagnosis delay, region of onset, age, gender, and comorbid frontotemporal dementia were performed to evaluate the functional and respiratory status of the patients at baseline and during disease progression – assessed using the global ALSFRS-R score and its respiratory subscore, and the predicted forced vital capacity (%FVC). A Cox regression controlling for the same variables was carried out to analyse survival.

**Results:** At baseline, 32/372 (8.60%) patients carried the *C9orf72* repeat expansion. We found that the *C9orf72* mutation is an independent risk factor for a faster %FVC decline ( $p = .001$ ) and shorter survival ( $p = .002$ ).

**Conclusions:** In ALS patients with *C9orf72* expansion, shorter survival probably derives from faster respiratory function decline. This finding may indicate a new pathogenic mechanism of *C9orf72* in ALS.

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## Risk indicators in families with abused children and young people: scoping review

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### ABSTRACT

**Introduction:** The promotion of rights and the protection of children implies involvement between state agencies and families in the evaluation processes, having developed in the child Welfare systems Internationally [1–3]. Although effective involvement is an essential component of the Help process [4], it presents continuous challenges for Professionals [5]. There is also an underlying tension between the regulatory role inherent in the protection system and the importance of involvement and contribution to develop the capacities of families producing better outcomes for children [6].

It is a challenge for families and professionals to deal with the duality of the relationship, given the expectations that workers engage in conflicting roles of supporting families, on the one hand, and ensuring the safety of children, on the