

POINTS TO REMEMBER FOR CLINICAL PRACTICE

Respiratory
pathologies

Obstructive Sleep Apnoea Syndrome (OSAS)

- The greater prevalence and severity of OSAS in people with DS than in the general population.
- In children, the prevalence is inversely proportional to age and 50% of cases are diagnosed before the age of 5.
- The clinical signs of OSAS are neither sensitive nor specific and cannot by themselves confirm or deny the diagnosis.
- The questionnaires tested in children with DS are less effective than in the general population; the better results obtained in adults with DS need to be confirmed.
- Nocturnal type 1 PSG is the reference test for diagnosing OSAS in DS.
- The consequences of OSAS in DS are multiple and the main systems affected are: the cardiovascular system, the central nervous system and metabolic functions.
- Preventing these consequences justifies systematic screening for OSAS by nocturnal type 1 PSG before the age of 4 years. In the absence of PSG, it is preferable to use alternative screening tests for OSAS rather than not performing objective tests at all.
- Given the pre-existing ID, the consequences of OSAS on language, particularly in very young children, justify screening by PSG as early as possible, and for some as early as 6 months of age.

- The consequences of OSAS on mood disorders and development of early dementia are important to consider in the adult with DS.
- Once a diagnosis has been made, treatable causes should be identified and included in a treatment algorithm, especially in cases of persistent OSAS after first-line treatment.
- At each stage, it is necessary to monitor the efficacy of the treatment with a PSG.
- In children with DS, the first-line treatment is adenotonsillectomy (AT) indicated in cases of moderate or severe OSAS (AHI $\geq 5/h$) confirmed by PSG and the presence of "significant" adenoidal and tonsillar hypertrophy. It is curative in 20-50% of cases.
- Mild forms of OSAS (AHI ≥ 2 and $< 5/h$) are amenable to a controlled trial of first-line drug treatment, the failure of which leads to surgical treatment.
- In case of failure of the first-line surgical or medical treatment (persistence of AHI $\geq 5/h$), CPAP will be prescribed as a second-line treatment.
- In adults with DS, CPAP is the first-line treatment for moderate to severe cases of OSAS (AHI $\geq 15/h$). It is effective with good feasibility, low complication rate, low failure rate and good compliance. In case of failure, other treatment options (orthodontic, surgical, medical) have not yet been adequately evaluated in DS, except for hypoglossal nerve stimulation which seems to be an effective technique in the medium term.

