prenato prenatal genetic screening specialist

Screening for microdeletions

Microdeletions are generally accidental and not inherited from parents *(de novo)*.

Detection rates vary from 87.5% to 100%.

Syndromes	Definition	Prevalence	Description	Management	PPV*[1]
22q11.2 deletion (DiGeorge syndrome)	Chromosomal abnormality caused by loss of a small fragment of chromosome 22	Approximately 1/2000 to 1/4000 live births (L.B.)	**Great variability in expression and severity.** Some people will have severe features while others will be diagnosed late due to a mild form. **<u>The main charecteristics</u>** -Heart defects - Facial dysmorphia - Delayed psychomotor development with/without intellectual disability - Behavioural disorder - Speech difficulties related to soft palate dysfunction - Hypocalcemia - Immunodeficiency	Multidisciplinary and variable management according to presentation and characteristics	89.3%
5p monosomy (Cri du Chat syndrome)	Chromosomal abnormality caused by partial or total loss of the short arm of chromosome 5	Approximately 1/15 000 to 1/45 000 L.B.; affects females more frequently than males	**Wide range or characteristics :** - Typical infant cry - Facial dysmorphia - Global neurodevelopmental disorder - Apnea episodes with cyanosis - Suckling difficulties in neonatal period - Small weight for gestational age / IUGR - Less frequent complications: cardiac, neurological, renal, other	Multidisciplinary management: treating professional, specialist physicians, paramedical professionals	66.7%
1p36 deletion	Chromosomal abnormality caused by partial loss of a segment of chromosome 1	Approximately 1/4000 to 1/10 000 L.B.	- Facial dysmorphia - Hypotonia - Developmental delay - Intellectual disability - Seizures - Heart defects - Absence or delay of language - IUGR	Multidisciplinary management: treating professional, specialist physicians, paramedical professionals	8.1%

Table created in June 2024 by A.Brousseau (RN) and C.Bascunana (G.C.)

*[1]PPV : Positive predictive value following a positive screening test

4p-monosomy (Wolf-Hirschhorn syndrome)	Chromosomal abnormality caused by loss of a segment of the short arm of chromosome 4	Approximately 1/20 000 to 1/50 000 L.B. Affects females more frequently than males	- IUGR and low weight gain at birth - Facial dysmorphia - Skeletal anomalies - Hypotonia - Severe developmental delay - Seizures - Cardiac, ophthalmological, auditory, dental anomalies	Multidisciplinary management: treating professional, specialist physicians, paramedical professionals	14.8%
Angelman Syndrome	Chromosomal abnormality caused by the alteration or absence of one or more gene(s) on chromosome 15	Approximately 1/12 000 à 1/20 000 L.B.	**Features always present** - Overall delay in acquisitions - Generally severe intellectual disability - Generally severe language disorder - Motor development disorder - Characteristic social behaviour: jovial, excessive laughter, agitated **Common manifestations** - Seizures - Reduced growth of the cranial perimeter resulting in microcephaly **Other features may be present less frequently**	Multidisciplinary management: treating professional, specialist physicians, paramedical professionals	20%
Prader-Willi syndrome	Chromosomal abnormality caused by the alteration of certain genes on chromosome 15	Approximately 1/20 000 to 1/25 000 L.B.	**Characteristics from birth to 2 years – Significant hypotonia** - Trouble sucking/swallowing - Delayed acquisition of language and walking - Respiratory infections - Speech and chewing problems - Characteristic facial features **Caractéristics after 2 years** - Hyperphagia (excessive appetite) leading to overweight and obesity. - Underdevelopment of the sexual organs - Moderate and variable intellectual disability - Learning difficulties and language disorders - Behavioural disorders (temper tantrums)	Occupational therapy, physiotherapy, diet, growth hormones, endocrinology and other medical and paramedical specialists	20%

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