

22 - Clinical features

Clinical features

820 The Maudsley® Prescribing Guidelines in Psychiatry CHAPTER 10 22q11.2 deletion syndrome

Clinical features 22q11.2 deletion syndrome (22q11.2DS), the most common microdeletion syndrome in humans, is a multisystem disorder with a heterogenous presentation which varies greatly in severity between affected individuals.¹ Prevalence is estimated at 1 in 2148 live births.^{1,2} The syndrome has been known by many names (including velocardiofacial syndrome and DiGeorge syndrome) in part because of its broad phenotypic range of clinical features (Table 10.6).

Table 10.6 Selected clinical features and risks in people with 22q11.2 deletion syndrome.^{1,3}

Clinical risks Cognitive and adaptive functioning Intellectual disabilities Deficits in adaptive functioning Endocrinological Hypocalcaemia and hypoparathyroidism Hypomagnesaemia Thyroid disease (usually hypothyroidism) Obesity and type 2 diabetes mellitus Gastroenterology General gastrointestinal symptoms (constipation, dysphagia) GERD Fatty liver Psychiatric disorders Anxiety Psychosis Autism spectrum disorder ADHD Cardiovascular Congenital cardiac defects Hypertension, arrhythmia, heart failure, aortic root dilation Genitourinary Congenital anomalies, renal cysts, renal failure Menstrual disorders (e.g. dysmenorrhoea) Neurology Seizures, often secondary (e.g. to hypocalcaemia) Early-onset Parkinson's disease Other motor disorders (e.g. dystonia, myoclonus) Sleep Sleep pattern disruptions Obstructive sleep apnoea Haematology Mild to moderate thrombocytopenia, mild cytopenias Immune thrombocytopenia Impaired haemostasis (e.g. epistaxis, menorrhagia) GERD, gastro-esophageal reflux disease.

Revision #1

Created 2026-01-04 20:17:44 UTC by Omar Ayman

Updated 2026-01-04 20:17:44 UTC by Omar Ayman