

Oral-facial-digital syndrome (OFDS) is an umbrella term for at least 10 apparently distinctive genetic disorders that are characterized by defects and flaws in the development of the structure of the oral cavity including the mouth, tongue, teeth, and jaw; the development of the facial structures including the head, eyes, and nose; and the fingers and toes (digits); along with differing degrees of mental retardation. The presentation of signs and symptoms is extremely varied, making diagnosis difficult. OFDS type I is the most common of all of these disorders, and it is quite rare. Each of the other types is extremely rare. All types of oral-facial-digital syndrome are rare, with type I being the least rare. The incidence of OFDS type I is thought to be between 1 per 50,000 births and 1 per 250,000 births. Diagnosis of OFD syndrome type I when suspected, may be confirmed by genetic testing. There are no specific tests at the present time for any of the other types. However, diagnosis is generally made on the basis of the clinical symptoms presented.