PRODUCT CATALOGUE

primeFISH Products

MICRODELETION PANEL

Discovering Life, Enriching Futures

DIAGEN BIOTECHNOLOGY Inc.

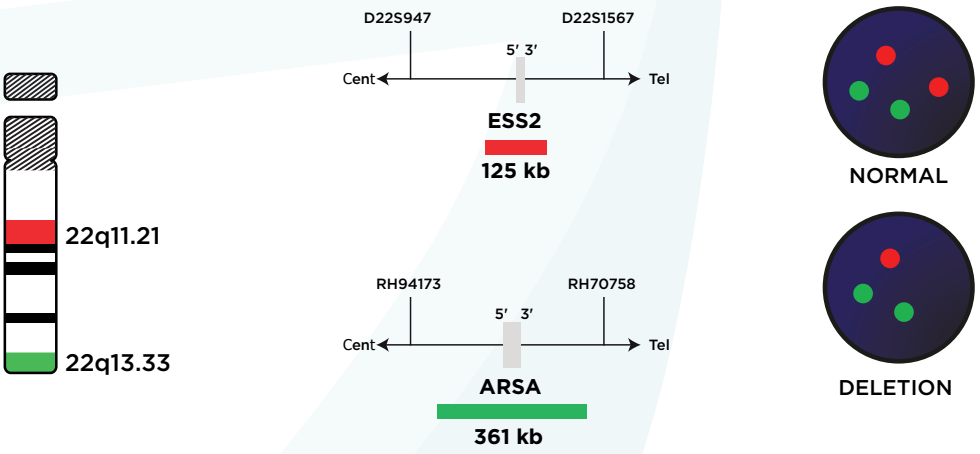
Stratech

MICRODELETION PANEL

17-019 Digeorge N25

22qll.2 deletion syndrome, alsa known as velocardio-facial syndrome (VCFS) and DiGeorge syndrome, is a genetic disorder caused by hemizygous microdeletions on chromosome 22qll.2. The population prevalence is l in 4000 births. The characteristic phenotype of dele tion of 22qll.2 includes cardiac defects, immunodeficiency, growth restriction, and cognitive deficits. 22qll.2 deletion usually occurs by meiotic non-allelic homologous recom-bination events between low copy repeats called LCR22 on chromosome 22qll.2.

MICRO**DELETION**



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References

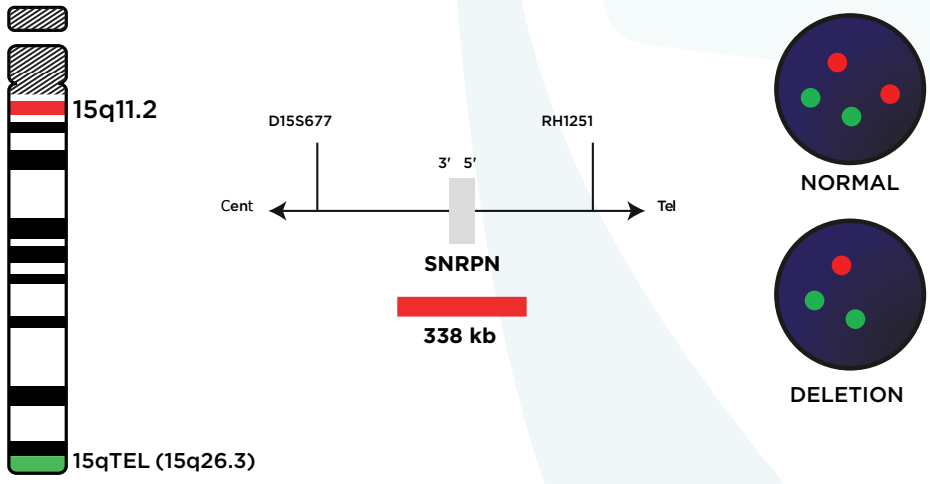
Michaelovsky E, et al. (2012) BMC Med Genet 13: 122
Morrow BE, et al. (2018) Am J Med Genet A 176: 2070-81.
Wilson HL et al., J Med Genet 2003;40(8):575-84

MICRODELETION PANEL

17-018-A PW/ANGELMAN

Prader-Willi syndrome (PWS) is a sporadic genetic disorder caused by genomic errors that disable paternally inherited genes in the PWS critical region on chromosome l5qll-ql3. Lack of expression of one or more of these genes results in different PWS phenotypes. There are three main genetic causes of PWS syndrome: it is caused by paternal 5-7 Mb deletion of the l5qll-ql3 region, uniparental disomy 15 from the maternal side, or defects in the PWS critical region. The SNRPN (small nuclear ribonucleoprotein polypeptide N, alsa known as PWCR) gene is located in the PWS region and has an important regulatory role on imprinted genes located on chromosome 15. The esti-mated prevalence of the disease ranges from l/15,000 to l/30,000 newborns. Clinically, PWS patients present with poor sucking and poor weight gain in infancy with a characteristic appearance, including hypotonia, mild mentol retardation, hypogonad ism, growth hormone deficiency leading to short stature, early childhood onset of hyperpha gia and obesity, characteristic appearance, and behavioral and sometimes psychiatric observations exhibits a pattern of symptoms.

MICRODELETION



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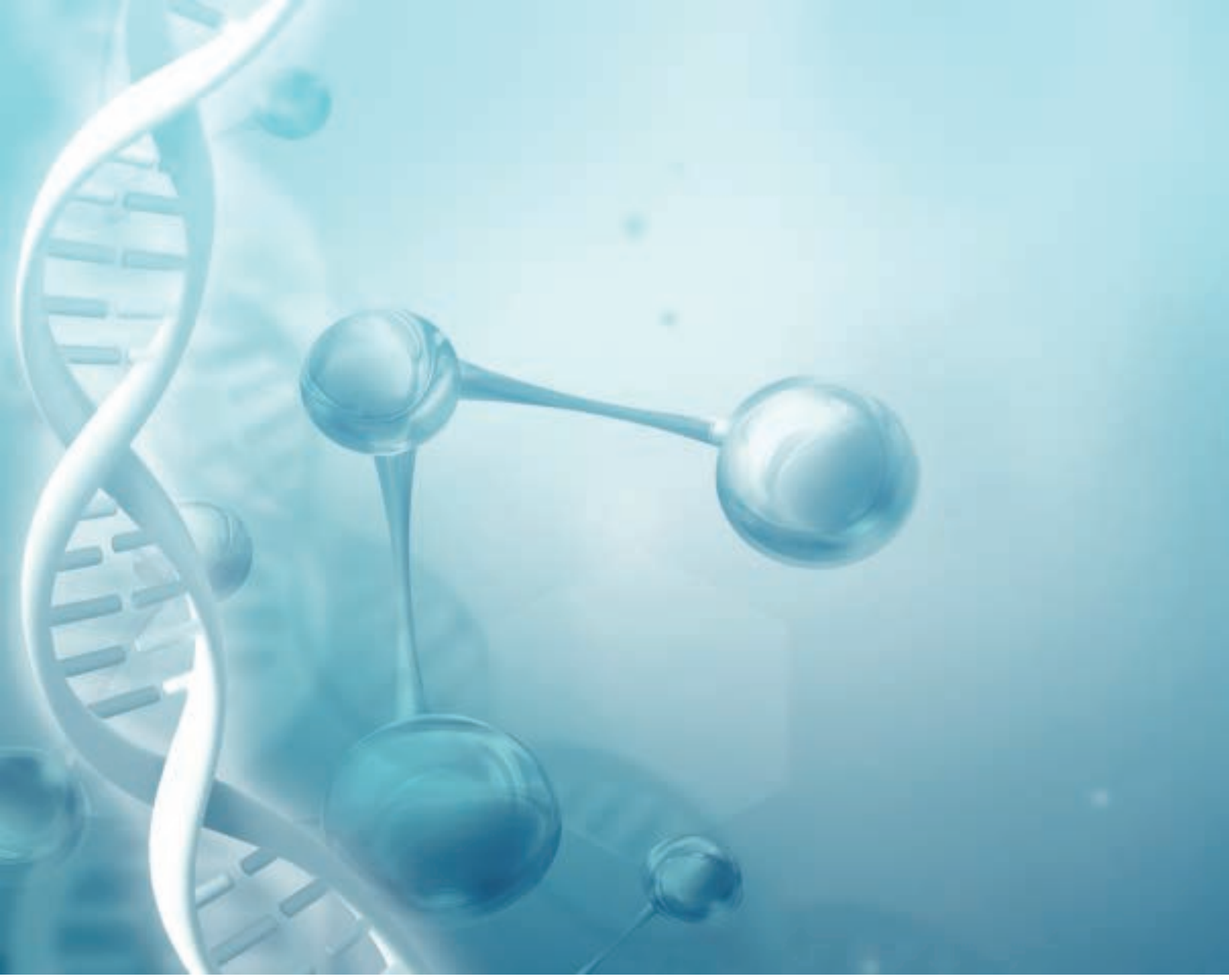
References

Reifenberger et al (1994) Am J Pathol 145:1175-1190
Louis et al (2016) Acta Neuropathol 131:803-820
Staedtke et al (2016) Trends Cancer 2:338-349



Stratech

www.stratech.co.uk

info@stratech.co.uk
+44 (0) 1638 782600
@stratech_uk
@stratech-scientific-ltd
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