CHONDRODYSPLASIA PUNCTATA

**Published Data**

**In-house data (19/35 positive)**

- CDP with distal phalangeal hypoplasia (males)
- CDP with raised 8-dehydrocholesterol and 8(9)-cholestenol
- CDP with no family history & no biochemical information
- RCDP with biochemical evidence of RCDP3
- RCDP with biochemical evidence of RCDP2
- RCDP with biochemical evidence of RCDP1

ARSE (75%)
EBP(*100%)
EFP (*54%)
AGPS (*100%)
GNPAT (*100%)
PEX7 (*100%)

\[ a = \text{Alkyl-Dihydroxyacetonephosphate Synthase deficiency} \]
\[ b = \text{Acyl-CoA:dihydroacetonephosphate acyltransferase deficiency} \]
\[ c = \text{Deficiency of plasmalogens in red blood cells, increased plasma concentration of phytanic acid and normal plasma concentration of very long chain fatty acids.} \]