

Figure S3. Venn diagram to show clinical overlap and diversity in VPS35L-, WASHC5-, and CCDC22-associated Ritscher-Schinzel syndrome (RSS).

Clinical manifestations reported in VPS35L-, WASHC5-, or CCDC22-associated RSS are summarized in this Venn diagram. The bold and italic text represent classical triad of RSS, also known as 3C (cranio-cerebello-cardiac) syndrome. Dyslipidemia is highlighted in bold red text because its pathomechanism was intensively studied in this report.